ABSTRACT

FINGERPRINT PATTERNS IN IDIOPATHIC MENTAL RETARDATION

By

Terry Jon Hassold

Fingerprint patterns from 1463 Caucasian control individuals (733 males, 730 females) and 868 non-specific mentally retarded individuals (537 males, 331 females) were examined to determine:

1. if differences with respect to fingerprint patterns existed between the two groups and
2. if such differences might be used to uncover previously undetected mental retardation syndromes.

Fingerprints were analyzed separately by sex and, for the retarded group, were subdivided on the basis of severity of retardation. The fingerprints of the control and retarded groups were compared with respect to total finger ridge count, pattern intensity, frequency distribution of the four digital patterns, and ten-finger patterns. Retarded individuals of both sexes showed an increased frequency of arches. Additionally, among the retarded males, there was an overall decrease in the frequency of whorls; among the retarded females, there was an increase in whorls and a decrease in ulnar loops.

Examination of ten-finger patterns led to the detection of 18 categories of such patterns in excess among mentally retarded individuals of either or both sexes. Forty-seven individuals from two of these categories (i.e., arches or radial loops on digit I) were further evaluated for possible similarities indicative of a mental retardation syndrome. Among these individuals, higher than expected frequencies of cryptorchidism (28.6% of the males) and autosomal chromosome abnormalities (8.5%) were noted. Two of the individuals with chromosome abnormalities had cytogenetic and clinical similarities consistent with that of a chromosomal syndrome.

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By

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To my parents, for their patience, guidance, and love

To Benita, for her love and support

To "the Boss", Herman M. Slatis, for his supervision and friendship

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INTRODUCTION

During the third and fourth months of human fetal development, epidermal ridge configurations are formed on the palmar and plantar surfaces of the body. These configurations will later be recognizable as the fingerprints, palmprints, and footprints, and are distinctive in several ways:

1. because of their early time of formation, they can act as sensitive indicators of developmental disruptions occurring early in fetal life
2. once formed, they are unaffected by environmental or aging factors, and retain the same morphology (except for size) for the lifetime of the individual
3. they show considerable variability; no two individuals possess the exact same set of configurations.

The study of these ridge configurations is known as dermatoglyphic analysis, and has come to play an increasingly important role in medical genetics. Many genetic disorders, particularly those associated with mental retardation, are now known to include characteristic dermatoglyphic abnormalities. In fact, for certain of these disorders it has been possible to formulate diagnostic indices based solely on dermatoglyphic discriminants. Such indices have been particularly useful in diagnosis of specific chromosomal abnormalities (e.g., trisomies 13, 18, and 21), although for each of these syndromes a definitive diagnosis can be achieved only through cytogenetic evaluation.

Most of the diagnostic indices that have been formulated have used both hand- and footprint patterns in developing appropriate discriminants. However, in one study (Slatis and Hassold, 1971) a comparison of fingerprint patterns between normal and Down's syndrome individuals led to the formulation of an index capable of diagnosing Down's syndrome with 95% accuracy. This degree of accuracy compares favorably with other, more complex, dermatoglyphic indices for the diagnosis of Down's syndrome.

The purpose of the present study was to determine the usefulness of such a fingerprint analysis as a screening technique in idiopathic mental retardation. Specifically, the ten finger patterns of control and retarded individuals were to be compared to determine if there exist, within the retarded population, any sub-groups with similar, non-normal fingerprint patterns. If such sub-groups were isolated, individuals within a group were to be examined with respect to family history, physical stigmata, and possible biochemical or chromosomal abnormalities. From such examinations, it was thought that previously undetected, mental-retardation syndromes might be uncovered.

REVIEW

Embryogenesis and classification of fingerprint patterns

Fingerprint patterns are known to develop in association with the volar pads (Mulvihill and Smith, 1969). The pads are aggregations of mesenchymal tissue which occur as elevations on each of the toe- and fingertips and on certain parts of the palm (beneath each of the digits and in the hypothenar area, the proximal part of the ulnar side of the palm). The volar pads are first observed at the sixth week of development, and reach their maximal growth between the tenth and twelth weeks (Cummins, 1929; Hale, 1952). As the volar pads begin to recede, the fingerprints are formed in the spaces previously occupied by the pads (Hirsch and Schweichel, 1973). According to several investigators, the type of fingerprint pattern is dependent on the shape of the volar pad which preceded it (Crawford, 1968; Penrose, 1968; Mulvihill and Smith, 1969; Hirsch and Schweichel, 1973; Okajima, 1975). The entire process of fingerprint pattern formation is completed between the 19th and 20th weeks of gestation (Cummins, 1929; Hale, 1952; Preus and Fraser, 1972). Once formed, fingerprint patterns retain the same morphology (except for size) for the life of the individual (Alter, 1967; Holt, 1968).

Fingerprint patterns are categorized on the basis of three anatomical features - the triradius, the type lines, and the pattern core (Figure 1). Type lines are ridges which immediately surround the

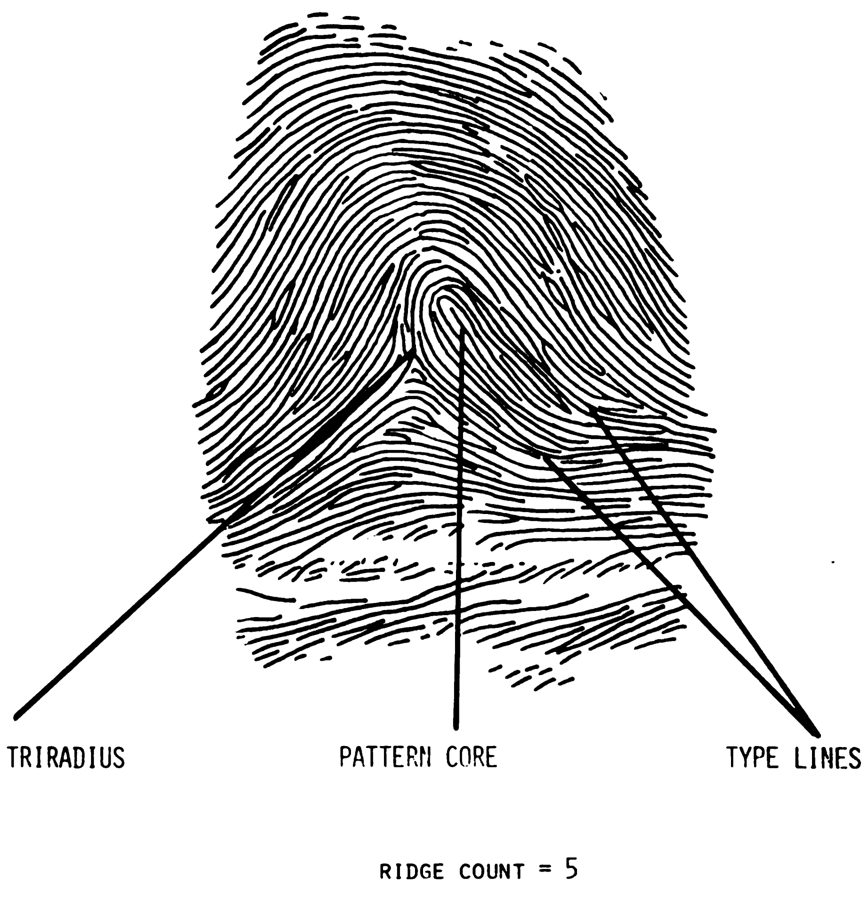

FIGURE I. Anatomical features of a typical fingerprint pattern

pattern area, thereby defining the limits of the pattern. The type lines are formed by the triradius, which is a junction of three ridges. The pattern core is the apparent center of the pattern area. The ridge count, a measurement made on fingerprint patterns, is defined as the number of ridges between the triradius and the pattern core, minus the ridges at the triradius and pattern core.

Based on these features, three different types of fingerprint patterns are recognized - arches, loops, and whorls. These different types of patterns are illustrated in Figure 2. An arch is a non-pattern in which the ridges flow from one side of the finger to the other. Two types of arches may be distinguished: simple arches, which do not have a triradius, and tented arches, with a triradius. By definition, arches always have a ridge count equal to zero. A loop is a pattern in which the ridges enter the pattern area from one side of the finger and curve back to exit on the same side of the finger. There are two types of loops: ulnar loops and radial loops. In the former case the loop opens toward the ulnar side of the hand; in the latter toward the radial. A loop has one triradius and one ridge count (equal to at least one). A whorl, the most complex type of fingerprint pattern, is characterized by a circular or elliptical shape with at least two triradii and two ridge counts. Rarely, whorls may have three triradii and, in such cases, three ridge counts.

Techniques for the analysis of fingerprint patterns

Early studies of fingerprint patterns were concerned with their inheritance and their usefulness in analyses of racial variations.

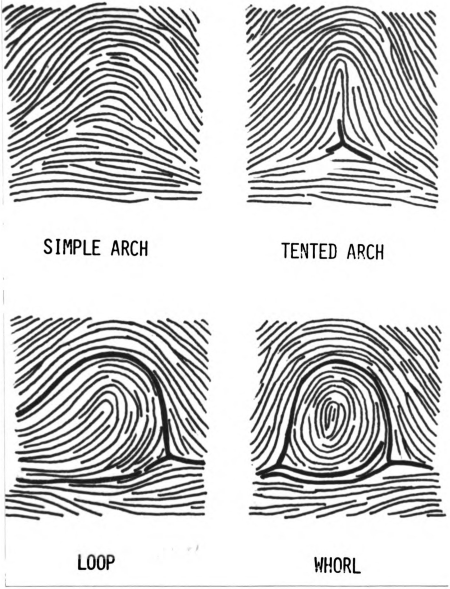

FIGURE 2. Different types of fingerprint patterns

However, with Cummin's discovery (1936) of palm- and fingerprint differences between Down's syndrome and normal individuals, the possible diagnostic value of dermatoglyphic analysis became apparent. Since that time, the majority of fingerprint analyses have been aimed at the detection of pattern abnormalities for a variety of genetic and environmental disorders. The analytical techniques used in these studies can be grouped into three general categories:

1. analysis of pattern type and intensity
2. analysis of ridge counts
3. analysis of the ten fingerprint patterns as one entity.

1. Analysis of pattern type and intensity

The simplest and most common method of study seen in analyses of fingerprint patterns involves an analysis of the frequency distribution of pattern types. For each of the ten fingers, the pattern type is recorded and the findings compared for the populations under study. Two extensions of this analysis are the arch-whorl index (Dankmeijer, 1934) and the pattern intensity index (Cummins and Midlo, 1935).

The arch-whorl index was designed to determine the relative simplicity of complexity of fingerprint patterns for a given population. For such purposes, the populations being examined are given a score. The score is arrived at by dividing the number of arches seen by the number of whorls, and multiplying by 100 (arch/whorl x 100). This index has been of little value in medical genetics because; 1. not all individuals within a population can be assigned a score (i.e., individuals with no whorls have a mathematically undefined score), 2. scores may be greatly exaggerated if the group being studied includes only a small number of individuals (i.e., if an individual within the group has all arches or whorls, the overall score may be artificially inflated or deflated), 3. the index gives no indication as to the distribution of whorls or arches on the individual digits.

Like the arch-whorl index, the pattern intensity index is intended to indicate the relative complexity of fingerprint patterns within a given population. Scores are determined by summing the number of triradii on the ten fingers for each individual (arch = 0, ulnar loop = 1, radial loop = 1, whorl = 2). Hence, pattern intensity scores normally range from 0 to 20 (except in the rare cases where whorls with three or more triradii are seen). Unlike the arch-whorl index, pattern intensity values can be assigned to any individual. However, similar drawbacks with respect to small groups and specific comparisons of pattern type distribution limit the applicability of this analysis for medical genetics.

2. Analysis of ridge counts

Ridge count analysis was first formally discussed by Bonnevie (1924). Like the arch-whorl and pattern intensity indices, it has been used to indicate the relative complexity of patterns in an individual or population. Ridge counts from each finger may be examined separately or summed to give a single value per individual. Holt (1968) distinguished between two types of summed values: the absolute ridge count, in which all ridge counts are summed (since whorls have two ridge counts, this value may involve summation of more than ten ridge counts), and the total finger ridge count, in which one ridge count per finger is included in the total value (in the case of whorls, only the larger of the two ridge counts is summed).

Of the two methods of ridge count analysis, the total finger ridge count is the one most often used. By itself, this analysis is of little value in medical genetics, because it includes no description of the pattern types and their distribution on the fingers. However, when used in combination with such a description, it adds considerable knowledge about the complexity of the patterns.

3. Analysis of the ten fingerprint patterns as one entity

In a comparison of fingerprint patterns of normal individuals, Down's syndrome individuals, and idiopathic mentally retarded individuals, Lu (1968) analyzed the ten fingerprint patterns from each individual as if they were one dermatoglyphic trait. Thus, an individual with ten ulnar loops was considered to have one pattern, an individual with nine ulnar loops and a whorl on digit I of the right hand a second pattern, and an individual with nine ulnar loops and a whorl on digit I of the left hand a third pattern. In this manner, Lu hoped to demonstrate that unusual combinations of patterns, as well as unusual patterns by themselves, could be useful in discriminating between normal and non-normal groups. While Lu's study was hindered by a small sample size, subsequent studies (Slatis and Hassold, 1971; Thompsen and Bandler, 1973) have shown the usefulness of this type of analysis. In fact, a diagnostic index for Down's syndrome based largely on Lu's analytic technique was shown to have a 95% accuracy rate (Slatis and Hassold, 1971).

Fingerprint abnormalities associated with mental retardation

Abnormalities of fingerprint patterns have been seen in association with a variety of mental retardation syndromes. For most the abnormalities are subtle and are reflected as slight changes in pattern frequencies from those of control individuals. However, for certain chromosomal abnormalities the differences are of diagnostic usefulness. In the following section, a review is given of fingerprint abnormalities seen in association with chromosomal abnormalities, mental retardation syndromes not associated with chromosomal syndromes, and idiopathic mental retardation.

Chromosomal abnormalities

Down's syndrome

Down's syndrome has been the most extensively studied mental retardation syndrome with respect to dermatoglyphic analysis. Cummins (1936, 1939, 1943) was the first to describe significant abnormalities in the fingerprint patterns of Down's individuals. He reported that, among the Down's individuals, there was an increased frequency of ulnar loops and a concomitant decrease in the frequency of all other patterns. Cummins also found that, in Down's syndrome, radial loops were most commonly seen on digit 4 and digit 5 (among normal individuals, radial loops are frequently seen on digit 3 but are found only rarely on digits 4 and 5. All of Cummins' findings have been confirmed by several investigators (Walker, 1957, 1958; Geipel, 1961; Beckman et al., 1962; Holt, 1964; Soltan and Clearwater, 1965; Smith et al., 1966; Lu, 1968; Fujita, 1969; Giovannuci and Bartolozzi, 1969; Greyerz-Gloor et al., 1969; Shiono, 1969; Bryant, 1970; Borgaonkar, 1971; Slatis and Hassold, 1971; Thompsen and Bandler, 1973; Deckers et al., 1973).

Holt (1951) determined that, in Down's syndrome, the total finger ridge count is significantly reduced when compared to the normal population (136.4 vs. 128.7). Additionally, Geipel (1967) and Holt (1964) reported that, among Down's individuals, the loops tend to be high and L-shaped. Similar findings were reported by Slatis and Hassold (1971), and were used in constructing a diagnostic index for Down's syndrome.

Studies of translocation Down's syndrome individuals have yielded results essentially the same as for the typical trisomy (Rosner and Ong, 1967; Soltan and Clearwater, 1965). Mosaic Down's individuals have shown pattern frequencies intermediate between normal and trisomic individuals (Loesch, 1974).

Fingerprint pattern abnormalities have been utilized in the development of several dermatoglyphic indices for the diagnosis of Down's syndrome. Most of these indices also have included palmar and/or plantar patterns to help to discriminate between the Down's and normal populations (Cummins and Platou, 1946; Cummins et al., 1950; Turpin and Lejeune, 1953; Walker, 1957, 1958; Beckman et al., 1965; Greyerz-Gloor et al., 1969; Reed, 1970; Reed et al., 1971; Borgaonker, 1970; Deckers et al., 1973). Two indices, however, have been based solely on fingerprint patterns (Lu, 1968; Slatis and Hassold, 1971).

Trisomy 13

Characteristic fingerprint abnormalities associated with Trisomy 13 include an increased frequency of arches and radial loops, with an accompanying decrease in whorls and ulnar loops (Uchida et al., 1962; Penrose, 1966; Yunis, 1966). Lazyuk et al. (1971) found that almost 80% of Trisomy 13 individuals in their sample had at least one arch, and Preus and Fraser (1972) reported that 25% have four or more arches. Only 3% of control individuals have four or more arches (Preus and Fraser, 1972). The occurrence of radial loops on digits other than digit 2 has also been reported to be increased in Trisomy 13 (Lazyuk, 1971).

Analyses of individuals with partial trisomy for the proximal segment of chromosome 13 (Escobar and Yunis, 1974), and for the distal segment of 13 (Escobar et al., 1974; Schinzel et al., 1976) have not yielded any abnormalities with respect to fingerprint patterns.

Trisomy 18

The most obvious fingerprint abnormality associated with Trisomy 18 is a dramatic increase in the frequency of arches. Ten arches are seen in approximately 40% of all individuals with Trisomy 18 (Schauman and Alter, 1976), and fewer than six arches is rare (Preus and Fraser, 1972). The frequency of ulnar loops and whorls is much reduced in Trisomy 18 (Uchida et al., 1962; Uchida and Soltan, 1963; Penrose, 1969). The overall frequency of radial loops is about the same as for the general population; however, these are seen in increased frequency on digits other than digit 2, particularly on digit 1 (Preus and Fraser, 1972).

Wolf-Hirschorn syndrome (4p-)

Dermatoglyphic abnormalities reported in association with short arm deletion of chromosome 4 include hypoplastic dermal ridges, an increased frequency of arches, and a reduced total finger ridge count (Miller et al., 1969; Guthrie et al, 1971).

Cri-du-chat syndrome (5p-)

Warburton and Miller (1967) reported an increase in whorls and total finger ridge count, and a decrease in ulnar loops, among individuals with 5p-. These results were supported by the findings of Miller et al. (1969) and Niebuhr (1971).

Trisomy 8

The only obvious fingerprint abnormality seen among 11 individuals with Trisomy 8 or Trisomy 8 mosaicism was an increase in the frequency or arches, which accounted for 40% of all patterns seen (Schauman et al., 1974; Jacobsen et al., 1974). Accompanying this increase was the expected decrease in total finger ridge count. One individual with trisomy for the short arm of chromosome 8 had radial loops bilaterally on digit I, a finding almost never seen in control individuals (Preus and Fraser, 1972).

Trisomy 9p

In Trisomy 9p, whorls have been reported to be reduced and arches increased in frequency in a small series of individuals (Schwanitz et al., 1974; Blank et al., 1975; Lurie et al., 1976). Of five individuals with a partial deletion for the short arm of chromosome 9 (the anti-syndrome of trisomy 9p), four had six whorls and the fifth individual had ten whorls, findings exactly the reverse of that for the trisomic state (Alfi et al., 1976).

Deletion of the long arm of 18 (18q-)

Characteristic fingerprint abnormalities associated with 18q- include an increase in whorls and total finger ridge counts. All twelve individuals examined by Mavalwala et al. (1970), Plato et al. (1971), and Schinzel et al. (1974) had at least four whorls. The overall percentage of whorls for these individuals was 68%.

Deletion of the long arm of 22 (22q-)

Studies of eight individuals with 22q- showed an overall increase in the frequency of whorls (Schindler and Warren, 1973). All but one individual had at least four whorls, and the overall frequency of whorls among the eight individuals was 60%.

Sex chromosome abnormalities

Fingerprint abnormalities associated with sex chromosome abnormalities are less striking than those for autosomal errors, and involve minor shifts in pattern frequencies. Studies of Turner's syndrome have revealed a modest reduction in arches, balanced by an increase in ulnar loops. Frequencies for whorls and radial loops are very nearly the same for Turner's and normal females (Holt, 1964; Borgaonkar, 1970). Unexpectedly, there is a significant increase in total finger ridge count among Turner's individuals (Holt, 1964; Alter, 1965; Penrose, 1967; Borgaonkar, 1970). This is not the result of an increase of more complex patterns in Turner's syndrome; rather, it reflects the fact that, in Turner's syndrome, the patterns tend to be larger and have a greater number of ridges than seen in normals. Holt (1964) found no obvious differences in fingerprint patterns among Turner's individuals with different chromosome constitutions (i.e., XO, mosaic XX/XO, isochromosome X).

Fingerprint abnormalities associated with Klinefelter's syndrome (XXY) include a slight increase in the frequency of arches and a decrease in the total finger ridge count (Hunter, 1968; Cushman and Soltan, 1969). Similar findings, but more pronounced, have been seen for XXYY individuals (Uchida et al., 1964; Alter et al., 1966).

A relatively small number of XXX females have been studied with respect to dermatoglyphic analysis. One study revealed a significant decrease in total finger ridge count in 23 XXX females, when compared to control females (Penrose, 1967).

Studies of XYY males have shown a moderate excess of arches among these individuals, with a decrease in total finger ridge count (Tsuboi and Nielsen, 1969; Malalwaka, 1970; Borgaonkar and Mules, 1970; Saldana-Garcia, 1973).

Studies of total finger ridge count among normal males and females, and males and females with sex chromosome abnormalities, have shown that an inverse relationship exists between the number of sex chromosomes present and the total finger ridge count (Penrose, 1967).

Specific mental retardation syndromes not associated with chromosome abnormalities

Smith-Lemli-Opitz syndrome

Smith, Lemli and Opitz (1964) described the first three patients with this syndrome, and noted that two of the three had 10 whorls each. Dallaire (1969) described the dermatoglyphic findings in five additional patients; one had 10 whorls, one nine, and a third four. Shiono and Kadowaki (1975) summarized dermatoglyphic findings of 24 individuals cited in the literature, and reported that 10 had six or more whorls.

De Lange syndrome

Smith (1966) studied the dermatoglyphics of 21 individuals with de Lange syndrome and reported an overall reduction in whorls, and an increase in radial loops (particularly on digits 2, 3 and 4). These findings were corroborated by a second study of nine de Lange individuals (Abraham and Russell, 1968).

Rubinstein-Taybi syndrome

Robinson et al. (1966) reported dermatoglyphic abnormalities in two individuals with Rubinstein-Taybi syndrome. The first had eight digital arches, and the second had two patterns (a radial loop and an ulnar loop) on one digit I. In a second study of 19 affected individuals, an overall increase in the frequency of arches was reported (Giroux and Miller, 1967). Additionally, two of the individuals in this study were observed to have two patterns on one digit I. In a third study of 17 individuals, an overall increase in arches was noted (Simpson and Brissenden, 1973).

Congenital Rubella

The most consistent fingerprint abnormality associated with congenital rubella is an overall increase in whorls and a decrease in all other patterns (Alter and Schulenberg, 1966; Smith and Menser, 1968; Smith et al., 1969, Smith and Menser, 1973). Smith et al. (1969) noted that the overall percentage of whorls among 100 individuals with congenital rubella was 42.4% compared with 22.8% among controls, a statistically significant difference. Achs et al. (1966) suggested that among individuals with congenital rubella, the frequency of radial loops on digits other than digit 2 was increased. This, however, has not been confirmed by other investigators (Alter and Schulenberg, 1966; Smith and Menser, 1968; Smith et al., 1969).

Idiopathic mental retardation

Only a few studies have been done which compare fingerprint patterns among normal and non-specific mentally retarded individuals. In an early study, Bonnevie (1927) examined fingerprint patterns of 280 normal and 280 mentally retarded individuals. No attempt was made to separate the groups by sex or to categorize the retarded group by diagnosis. Bonnevie noted the following differences in the retarded group when compared to the controls:

1. an increased frequency of arches on digit 3 (.09 compared with .07)*
2. an increased frequency of large loops (not specified as ulnar or radial) on digit 1 (.36 vs. .28), digit 2 (.20 vs. .12), digit 3 (.38 vs. .28), and digit 4 (.25 vs. .21)
3. a decreased frequency of whorls on digit 2 (.20 vs. .25) and digit 3 (.08 vs. .13).

Blumel and Poll (1928) also found a decrease in the frequency of whorls among the retarded individuals they examined. However,

* values were extracted from graphs composed by Bonnevie and are only approximate

as in the case of Bonnevie's study, their retarded group has been described as "poorly defined" (Alter, 1966).

Alter (1966) analyzed dermatoglyphic patterns of 200 control and 376 non-specific mentally retarded individuals. Males and females of both groups were examined separately. Additionally, the retarded group was screened to remove from the study any individuals with known genetic or environmental causes for their retardation. No obvious differences with respect to fingerprint patterns were seen between the control and retarded individuals (Table 1).

Lu (1968) examined fingerprint patterns of 299 normal individuals (143 males and 156 females) and 281 non-Down's mentally retarded individuals (170 males and 111 females). Males and females were not analyzed separately. For each individual, the ten digital patterns were considered as a single entity, or "ten-finger pattern". Ten-finger patterns as well as frequencies of patterns on the individual digits, were compared between the control and retarded groups. Lu observed that, among the retarded group, ulnar loops were reduced on all fingers except digit 2 of the right hand, whorls were increased on all fingers, and arches were increased on digit 1 of both hands and reduced on seven of the eight remaining digits. The frequency of radial loops was not noticeably different between the two groups. Overall frequencies of ulnar loops, whorls, radial loops, and arches from Lu's study are shown in Table 2.

From the 281 retarded individuals, 151 ten-finger patterns were seen that were not observed among the control population. Conversely,

TABLE I. Overall frequencies of ulnar loops, whorls, radial loops and arches among 100 control males, 215 mentally retarded males, 100 control females and 163 mentally retarded females

MALES

	Control	Mentally retarded
Ulnar loop	.587	.584
Whorl	.300	.305
Radial loop	.042	.042
Arch	.071	.069

FEMALES

	Control	Mentally retarded
Ulnar loop	.603	.587
Whorl	.274	.304
Radial loop	.041	.026
Arch	.082	.082

TABLE 2. Overall frequencies of ulnar loops, whorls, radial loops, and arches among 299 control and 281 non-Down's mentally retarded individuals (from Lu, 1968)

	Control	Mentally retarded
Ulnar loop	.641	.600
Whorl	.235	.300
Radial loop	.049	.052
Arch	.074	.049

176 ten-finger patterns were found only in the 299 control individuals. However, for most of these ten-finger patterns, only a single control or retarded individual was observed. Thus, although Lu attempted to discriminate between those patterns which were indicative of the retarded group and those indicative of the control population, the effects of chance on his findings must be considered. However, one type of ten-finger pattern was seen to be greatly increased among the retarded group. Twenty-five retarded individuals (.089 of the total) and only 12 normal individuals (.040) were seen to have ten ulnar loops.

Saksena and Mathur (1974) examined the dermatoglyphics of 200 normal and 50 non-Down's mentally retarded individuals. No indication was given as to the number of individuals of each sex examined. Significant findings among the retarded group included:

1. an increase in whorls on digits 3, 4 and 5
2. an increase in arches on digits 1, 2 and 3

TABLE 3. Overall frequencies of ulnar loops, whorls, radial loops, and arches in 200 control and 50 non-Down's mentally retarded individuals (from Saksena and Mathur, 1974)

	Control	Mentally Retarded
Ulnar loop	.609	.380
Whorl	.330	.530
Radial loop	.020	.014
Arch	.042	.076

3. a decrease in ulnar loops on all digits
4. a decrease in radial loops on digits 2 and 3.

Overall frequencies of the four types of digital patterns from Saksena and Mathur's study is shown in Table 3.

Singh (1976) compared 53 non-specific mentally retarded males and 500 control males with respect to 15 dermatoglyphic features. Among the features studied were the ten individual ridge counts, overall frequency of ulnar loops, overall frequency of radial loops, frequency of two patterns on a single finger, total number of triradii on the ten fingers, and pattern asymmetry. A multivariate analysis of these 15 features was used in assigning two canonical variates to each of the groups. The variates were used in constructing a scatter diagram, on which the means of the two groups were examined to determine whether or not the distance between them was statistically significant. No such difference was observed.

It is difficult to compare these various studies of idiopathic mental retardation for two reasons:

1. for most of the studies, little information was given with respect to the make-up of the populations (i.e., severity of retardation, type of retardation, sex distribution, and racial distribution), and
2. in only two of the studies (Lu, 1968; Saksena and Mathur, 1974) were data presented with respect to the frequency distribution of the four fingerprint pattern types; it is interesting to note that in both of these studies, the retarded groups had a reduced frequency of ulnar loops, an increased frequency of whorls and arches, and about the same frequency of radial loops as the normal individuals.

In the present study, it was possible to improve upon these earlier studies in two ways: by increasing the sample size, and by better controlling the retarded group with respect to sex, race, and severity and type of retardation.

MATERIALS AND METHODS

Collection of data

Fingerprint patterns from 868 Caucasian mentally retarded individuals were collected from the Oakdale Center for Developmental Disabilities, Lapeer, Michigan, and the Coldwater State Home and Training School, Coldwater, Michigan. Prior to the collection of the prints, consent forms had been obtained from either the parents or legal guardians of approximately 600 of these individuals. The consent form used in obtaining this permission is shown in appendix I; addresses of the parents or legal guardians had been obtained from the institutions and the parents or guardians were then contacted by mail. Prints from the remaining individuals were collected from 1970-1972, prior to the time when consent forms were required by the institutions. Prints were obtained by applying sensitizing fluid to the digits and rolling the finger on photographic paper. Applicators, sensitizing fluid, and paper were obtained from the Faurot Company, New York City, New York. Only mentally retarded individuals with no known genetic or environmental cause for their retardation were included in the sample. Because of previously reported differences between males and females (Cummins and Midlo, 1943; Holt, 1968), the 868 individuals were separated into groups of 537 males and 331 females. Each sex was then divided into four groups on the basis of

TABLE 4. Division of the 868 retarded individuals by sex and level of retardation

Level of Retardation	Males	Females
Profound	135	102
Severe	143	70
Moderate	138	105
Mild	121	54

I.Q. scores: profound retardation (I.Q.=0-19), severe retardation (I.Q.=20-35), moderate retardation (I.Q.=36-51), and mild retardation (I.Q.=52-67). Thus, the 868 retarded individuals were divided into eight groups, as shown in Table 4.

Fingerprint patterns from 1500 control individuals (750 of each sex) were obtained from the Michigan State Police station at East Lansing, Michigan, and the Michigan State University School of Criminal Justice, East Lansing, Michigan. Patterns from 17 males and 20 females were subsequently discarded because of scarring, amputation, or smudged prints, leaving a total of 733 control males and 730 control females.

Analysis of fingerprints

Eight hundred individuals were arbitrarily selected for a comparison of quantitative dermatoglyphic traits between the retarded and control groups. Individuals selected included 200 control males, 200 control females, and 50 individuals from each of the eight retarded groups. For each individual, two quantitative measurements were made:

1. total finger ridge count; individual ridge counts were made according to the guidelines of Penrose (1968) and the highest ridge counts from each of the ten digits were summed to give the total ridge count
2. pattern intensity; the total number of triradii on the ten digits.

Digital patterns from all control and retarded individuals were analyzed and classified as either ulnar loops, whorls, radial loops, or arches, in accordance with Penrose (1968). The ten digital patterns of each individual were then considered together as a single dermatoglyphic pattern, or ten-finger pattern (Lu, 1968).

Physical examination

Forty-seven retarded individuals with similar, non-normal thumbprint patterns were examined with respect to physical stigmata, family, gestational, and developmental histories, and possible biochemical or chromosomal abnormalities. Records on file at the institutions were used to review previous physical examinations, histories, and urinalyses of the 47 individuals.

Physical examinations of 27 of the 47 individuals were carried out by staff physicians of the two institutions; the remainder were examined by the author and at least one other individual familiar with medical genetics. Two forms were used in describing physical stigmata of the individuals; the one more commonly used is shown in Appendix I.

Cytogenetic evaluation of the 47 individuals was carried out at the Hawthorne Center, Northville, Michigan. Four individuals with abnormal findings were referred to the Human Cytogenetics Laboratory at Michigan State University for confirmation of the abnormality and detailed cytogenetic examination. For each individual, two micro- and two macro-blood cultures were processed. Microcultures were established by adding .25-.50 ml. of whole blood to Gibco IA microculture tubes. Macrocultures were established by adding 1-2 ml. serum to flasks containing Gibco IA medium and supplimented with 15% fetal calf serum. Cultures were incubated at 37 degrees Centigrade for 68 hours, at which time colchemid (.5 mg/ml) was added to the cultures to arrest them at metaphase. After two hours, cultures were removed from incubation and harvested. The harvesting procedure included hypotonic treatment (.075 M KCl) and three changes of fixative (3:1 methanol: acetic acid). Slides were prepared by spreading 3-4 drops of culture material onto slides pre-cleaned in 95% methanol and flame drying.

Preliminary cytogenetic evaluation was carried out using giemsa: trypsin banded metaphase preparations. One to seven day old slides were incubated at 65° C for 12-24 hours. Slides were then immersed in phosphate buffer (0.025 M KH_2PO_4; pH 6.8) for 15 seconds to 5 minutes, and subsequently treated with trypsin (1 ml 0.1% trypsin: 8 ml methanol: 36.4 ml 0.025 M KH_2PO_4; pH 6.8) for 15 seconds to 5 minutes. Photographs of appropriate metaphases were taken using a Zeiss Photomicroscope II, using Pan-X film. Prints from the negatives were developed using Kodabromide F-2 paper.

Additional slides were analyzed using a sequential quinacrine to centromeric banding techniques (Caspersson, 1971). Fresh to 2 day old slides were stained in a solution of 0.5% equeous quinacrine dihydrochloride plus a small amount of quinacrine mustard (less than 0.1 mg/ml) for 5-10 minutes. Slides were rinsed for one minute in running tap water, and for 45 seconds in phosphate buffer (32 ml 0.1 M citric acid: 50 ml 0.2 M Na_2HPO_4 , pH 5.6), and mounted in the buffer. Metaphases were examined under a Zeiss Photomicroscope II, equipped with a HBO-200 mercury lamp, and using barrier filters 53/44. Photographs of good quality metaphases were taken using Kodak Tri-X film; prints from negatives were made with Kodabromide F-4 paper.

Oil was removed from the Q-banded slides by treatment with xylene, followed by three rinses in ethanol. Protocol for the centromeric banding was as follows: slides were treated in 0.2 N HCl for 20 minutes, rinsed in 2xSSC briefly, treated with saturated acqueous barium hydroxide for 5-7 minutes, and incubated at 65°C in 2xSSC for 12-24 hours. Slides were then stained in 5% giemsa. Photographs of metaphases previously analyzed with Q-banding were made on a Zeiss Photomicroscope II using Pan-X film. Prints from negatives were made using Kodabromide F-4 paper.

RESULTS

Analysis of fingerprint patterns was carried out in two steps. First, a study of quantitative traits was performed to determine if any differences could be detected between the control and retarded groups. Traits examined included total finger ridge count, pattern intensity, and overall frequency distribution of the four pattern types. Secondly, ten-finger patterns were analyzed to determine if differences existed between the control and retarded groups. In this analysis, however, the emphasis was not on the characterization of differences, but was placed on the determination of which retarded individuals should be examined for possible identification of new mental retardation syndromes.

Total finger ridge count

Findings with respect to total finger ridge count are represented graphically in Figures 3 and 4. For both sexes, the mean value for control individuals was higher than the overall mean for retarded individuals. Among the males, the mean values were 138.26±51.00 and 124.77±45.56 for the control and retarded individuals, respectively. Corresponding values for the females were 125.46±47.02 and 121.68±43.36. As can be seen, the difference between the control and retarded groups was greater among the males than the females. Additionally, examination

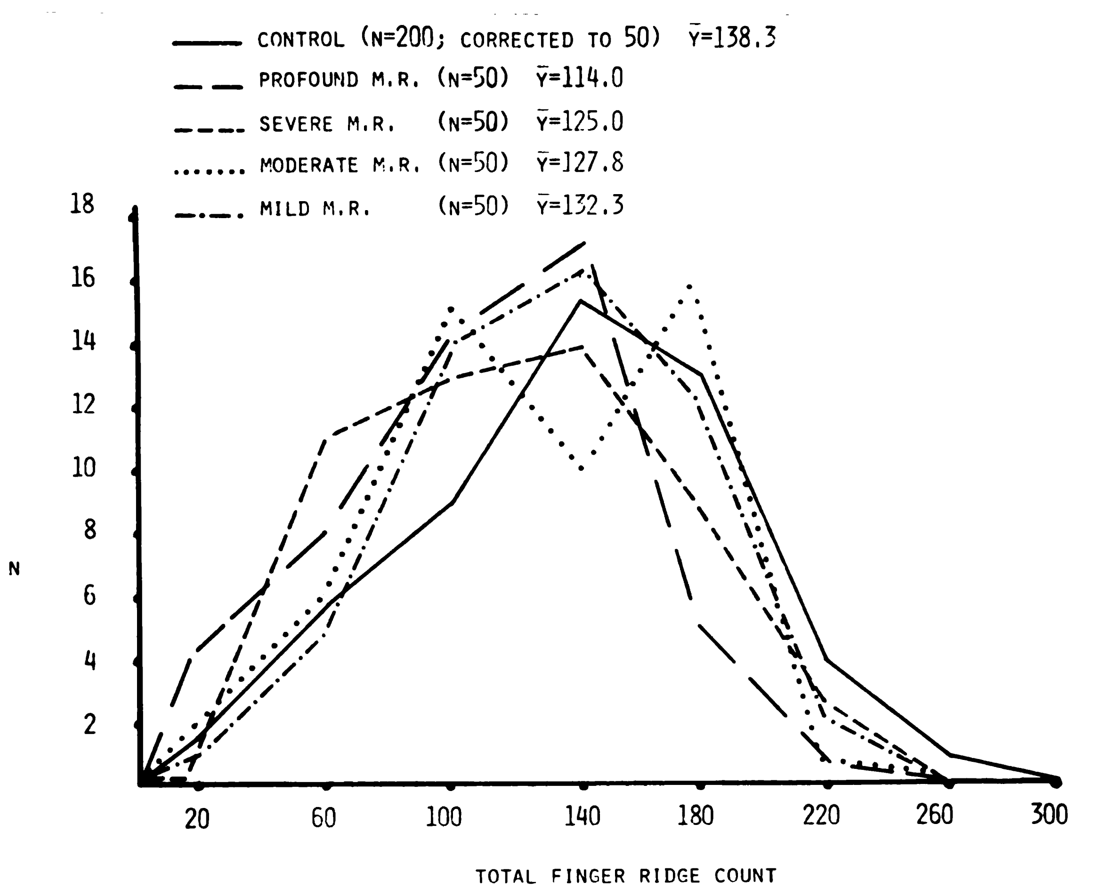

FIGURE 3. Total finger ridge counts in control and mentally retarded males

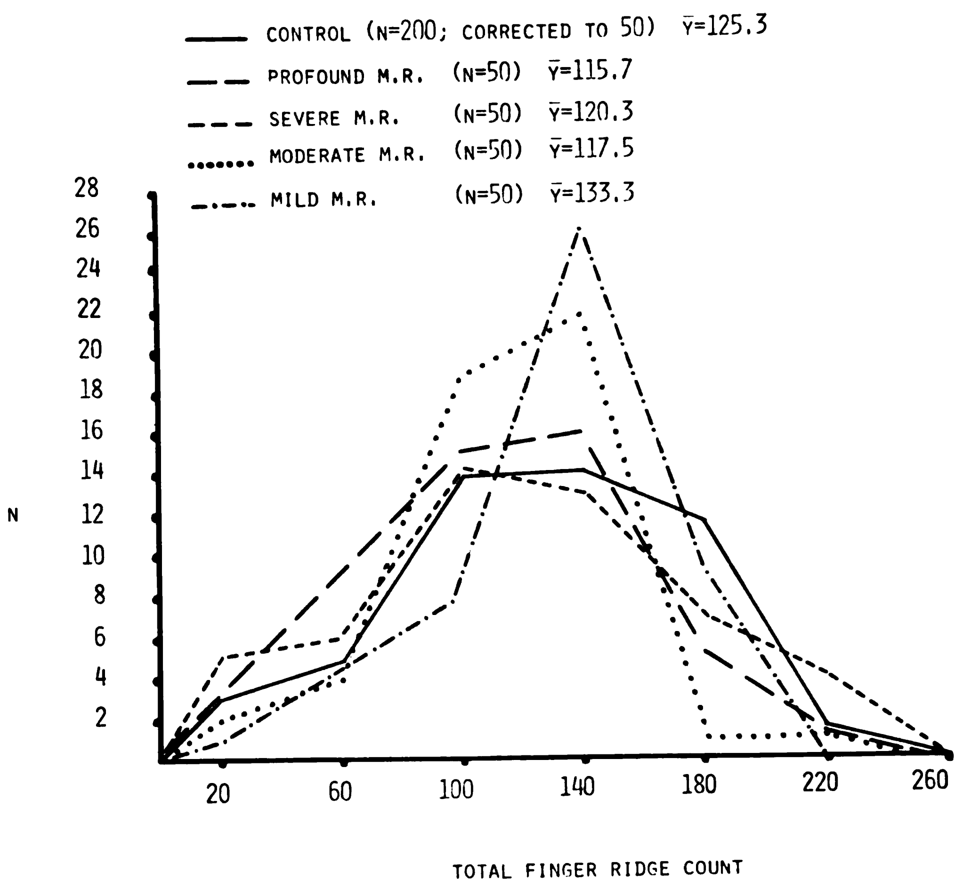

FIGURE 4. Total finger ridge counts in control and mentally retarded females

of the eight retarded groups revealed that, for each sex, the lowest mean values were found in the profoundly retarded groups, and the highest values were seen in the mildly retarded groups.

Two analyses of variance (one for each sex) were carried out to determine if significant mean differences existed between the control and retarded groups. Results are shown in Tables 5 and 6. Additionally, results of specific comparisons made using orthogonal contrasts are summarized in these tables. Results from these analyses reveal that:

1. among males the mean difference between the control and total retarded group was statistically significant at $p=.05$; however, the mean difference between the mildly retarded group and the other three retarded groups did not reach significance
2. among females the difference in means between the control and total retarded groups was not significant at $p=.05$; nor was the difference between the mildly retarded group and the other retarded groups.

Pattern intensity

Findings with respect to pattern intensity are represented graphically in Figures 5 and 6. No striking differences in mean values were detected between the retarded and control groups. Among the males, the mean for the control group was 12.44 ± 3.59, slightly higher than that for the retarded group, 12.28 ± 1.37. The reverse was true for the females; the control group had a mean value of 11.86 ± 1.38, and the retarded group a mean of 11.96 ± 5.10. As was the case for

TABLE 5. Analysis of variance to determine if significant mean differences exist between normal and retarded males with respect to total finger ridge count

Model = $Y_{ij} = \mu + \alpha_i + E_{(i)j}$ where

α_1 = control α_2 = profound retardation α_3 = severe retardation

α_4 = moderate retardation α_5 = mild retardation

ANOVA table

Source of variation	d.f.	SS	MS	f-ratio
I.Q. levels	4	27226	6806.5	2.917*
Error	395	921618	2333.2	
Total	399	948844		

$f_{.05,4,395} = 2.41$

Orthogonal Contrasts

	α_1	α_2	α_3	α_4	α_5
1. control vs. m.r.	+4	-1	-1	-1	-1
2. mild m.r. vs. remaining m.r.	0	-1	-1	-1	+3

1. $\Delta_1 = 13.485 \pm 9.48$ * for $\alpha = .05$

2. $\Delta_2 = 10.040 \pm 23.22$ for $\alpha = .05$

* significant at p = .05

TABLE 6. Analysis of variance to determine if significant mean differences exist between normal and retarded females with respect to total finger ridge count

Model = $Y_{ij} = \mu + \alpha_i + E_{(i)j}$ where

α_1 = control α_2 = profound retardation α_3 = severe retardation

α_4 = moderate retardation α_5 = mild retardation

ANOVA table

Source of variation				
I.Q. levels	4	10972.1	2743.03	1.344
Error	395	806175.9	2040.95	
Total	399	817148.0		

$f_{.05,4.395} = 2.41$

Orthogonal contrasts

	α_1	α_2	α_3	α_4	α_5
1. control vs. m.r.	+4	-1	-1	-1	-1
2. mild m.r. vs. remaining m.r.	0	-1	-1	-1	+3

1. $\Delta_1 = 3.78 \pm 8.87$ for $\alpha = .05$

2. $\Delta_2 = 15.49 \pm 21.72$ for $\alpha = .05$

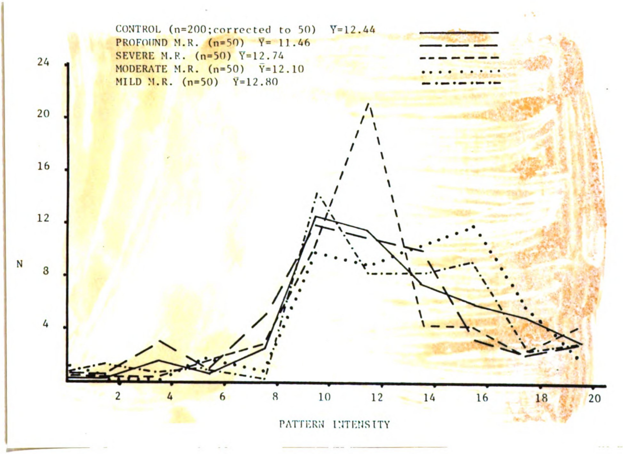

FIGURE 5. Pattern intensity in control and mentally retarded males

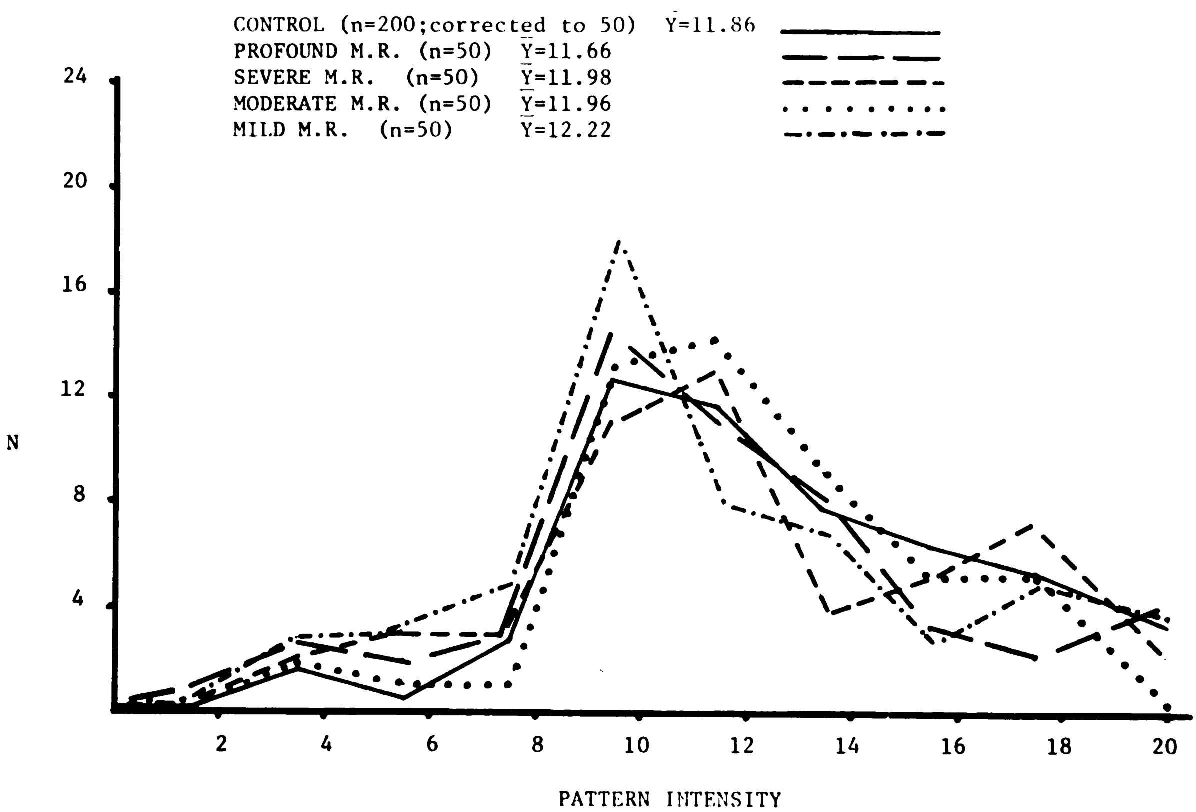

FIGURE 6. Pattern intensity in control and mentally retarded females

total finger ridge count, examination of the eight retarded groups revealed that, for each sex, the lowest mean value was seen in the profoundly retarded group and the highest value in the mildly retarded group.

Visual examination of the pattern intensity distributions showed them to be positively skewed (Figures 5 and 6). Thus, non-parametric tests were used in testing the hypothesis that significant differences in means existed between the various groups. Two Kruskal-Wallis tests were carried out, one for each sex. Data and results are shown in Tables 7 and 8. Differences were not significant for either sex (at p=.05).

Frequency distribution of the fingerprint patterns

Distribution of the four fingerprint types (ulnar loops, whorls, radial loops, and arches) among control males, control females, and the eight retarded groups is shown in Tables 9, 10 and 11. Tables 9 and 10 detail the distribution of the patterns on the individuals digits; Table 11 shows the overall distribution of the patterns. Several findings were seen to be common to the control groups of both sexes:

1. ulnar loops were the most commonly seen patterns (males=.593; females=.669); these were observed most frequently on digits 3 and 5 and were seen least frequently on digit 2,
2. whorls were the second most commonly observed patterns (males= .310; females=.219); together with ulnar loops, these two

TABLE 7. Kruskal-Wallis test to determine if significant mean differences exist between control and retarded males with respect to pattern intensity

Data-

		Control		Profound		Severe		Moderate		Mild	
Y	Rank(r)	n	(r)(n)	n	(r)(n)	n	(r)(n)	n	(r)(n)	n	(r)(n)
0	1		0	0	0	1	1	0	0	0	0
1	2	0	0	0	0	0	0	0	0	1	2
2		0	0	0	0	0	0	0	0	0	0
3	3.5	1	3.5	1	3.5	0	0	0	0	0	0
4	7.5	4	30	2	15	0	0	0	0	0	0
5	11.5	0	0	0	0	0	0	2	23	0	0
6	15.5	2	31	1	15.5	2	31	0	0	1	15.5
7	23.5	6	141	2	47	1	23.5	1	23.5	0	0
8	33	4	132	3	99	0	0	2	66	0	0
9	52.5	18	945	3	157.5	2	105	3	157.5	4	210
10	101	33	3333	9	909	8	808	7	707	10	1010
11	159	24	3816	6	954	7	1113	7	1113	5	795
12	206	22	4532	5	1030	1	206	14	2884	3	618
13	242.5	13	3152.5	5	1212.5	5	1212.5	1	242.5	4	970
14	273	17	4641	5	1365	4	1092	3	819	4	1092
15	306	11	3366	2	612	9	2754	4	1224	7	2142
16	331.5	13	4300.5	1	331.5	2	663	0	0	2	663
17	350	10	3500	1	350	4	1400	0	0	4	1400
18	368	10	3680	1	368	2	736	2	736	2	736
19	382.5	7	2677.5	1	382.5	1	382.5	1	382.5	2	765
20	394.5	5	1972.5	2	789	1	394.5	3	1183.5	1	394.5
$n_i=$		200		50		50		50		50	
$\Sigma r_i=$			40262.5		8641		10922		9561.5		10813

TABLE 7 (continued)

$$H = \left\{ \left[12/(\Sigma n)(\Sigma n+1) \right] \left[\Sigma(\Sigma r_i)^2/n_i \right] \right\} -3(\Sigma n+1)$$

$$=5.121$$

correction for ties:

$$D = 1- \quad T_j/(\Sigma n-1)(\Sigma n)(\Sigma n+1)$$

$$=.99$$

$$H_{corrected}=5.121/.99$$

$$=\underline{5.173}$$

Chi $square_{.05,4}=\underline{9.488}$

TABLE 8. Kruskal-Wallis test to determine if mean differences exist between normal and retarded females with respect to pattern intensity

Data-

		Control		Profound		Severe		Moderate		Mild	
Y	Rank(r)	n	(r)(n)	n	(r)(n)	n	(r)(n)	n	(r)(n)	n	(r)(n)
0		0	0	0	0	0	0	0	0	0	0
1	1.5	1	1.5	1	1.5	0	0	0	0	0	0
2		0	0	0	0	0	0	0	0	0	0
3	5.5	3	16.5	0	0	1	5.5	2	11	0	0
4	12	4	48	2	24	1	12	0	0	0	0
5	18.5	3	55.5	1	18.5	0	0	1	18.5	1	18.5
6	26	5	130	1	26	3	78	0	0	0	0
7	33.5	3	100.5	1	33.5	0	0	0	0	2	67
8	44	7	308	2	88	3	132	1	44	2	88
9	63.5	13	825.5	1	63.5	5	317.5	2	127	3	190.5
10	113.5	31	3518.5	13	1475.5	6	681	11	1248.5	15	1702.5
11	172	26	4472	4	688	5	860	3	516	3	516
12	219.5	23	5048.5	7	1536.5	8	1756	11	2414.5	5	1097.5
13	263	10	4997	1	263	3	789	6	1578	4	1052
14	295	17	5015	7	2065	1	295	3	885	3	885
15	320.5	14	4487	1	320.5	1	320.5	2	641	2	641
16	340	0	3060	2	680	4	1360	3	1020	1	340
17	358.5	10	3585	0	0	3	1075.5	2	717	3	1075.5
18	376	6	2256	2	752	4	1504	3	1128	2	752
19	387.5	3	1162.5	1	387.5	1	287.5	0	0	1	387.5
20	395.5	3	1186.5	3	1186.5	1	395.5	0	0	3	1186.5
		200		50		50		50		50	
Σr_i =			40273.5		9609.5		9969		10348.5		9999.5

TABLE 8 (continued)

$$H = \left\{\left[12/(\Sigma n)(\Sigma n+1)\right]\left[\Sigma(\Sigma r_i)/n_i\right]\right\} -3 \quad (\Sigma n+1)$$

$$= .223$$

correction for ties:

$$D = 1 - T_j/(\Sigma n-1)(\Sigma n)(\Sigma n+1)$$

$$= .989$$

$H_{corrected}$ = <u>.225</u>

Chi square$_{.05,4}$ = <u>9.488</u>

TABLE 9. Frequency of occurrence of digital patterns among males (733 control, 135 profoundly retarded, 143 severely retarded, 138 moderately retarded, 121 mildly retarded)

	Right Hand 1	2	3	4	5	Left Hand 1	2	3	4	5
CONTROL										
ulnar loop	370	206	542	341	562	468	267	546	445	613
	.505	.281	.740	.465	.767	.638	.364	.731	.607	.836
whorl	352	263	135	372	160	243	239	121	278	110
	.480	.359	.184	.508	.218	.328	.326	.165	.379	.150
radial loop	0	181	11	11	1	1	159	17	1	0
	.000	.247	.015	.015	.001	.001	.217	.023	.001	.000
arch	11	83	45	9	10	21	68	59	9	10
	.015	.113	.061	.012	.014	.029	.093	.080	.012	.014
PROFOUND										
ulnar loop	76	54	95	70	101	84	54	89	79	111
	.563	.400	.704	.519	.748	.622	.400	.659	.585	.822
whorl	51	44	26	62	27	40	40	30	48	19
	.378	.326	.193	.460	.200	.296	.296	.222	.356	.141
radial loop	1	21	2	0	3	0	24	5	0	0
	.000	.156	.015	.000	.022	.000	.178	.037	.000	.000
arch	8	16	12	3	4	11	17	11	8	5
	.059	.119	.089	.022	.030	.081	.126	.081	.059	.037

TABLE 9 (continued)

	Right Hand					Left Hand				
	1	2	3	4	5	1	2	3	4	5
SEVERE										
ulnar loop	78	38	95	51	103	95	54	87	78	111
	.545	.266	.664	.357	.720	.664	.378	.608	.545	.776
whorl	56	50	33	85	33	36	44	28	56	27
	.392	.350	.231	.594	.231	.252	.308	.196	.392	.189
radial loop	2	30	4	2	1	0	26	10	2	0
	.014	.210	.028	.014	.007	.000	.182	.070	.014	.000
arch	7	25	11	5	5	12	19	18	7	5
	.049	.175	.077	.035	.042	.084	.133	.125	.049	.035
MODERATE										
ulnar loop	78	41	95	73	115	88	53	95	94	120
	.565	.297	.688	.529	.833	.638	.384	.688	.681	.870
whorl	54	40	25	59	20	41	36	27	42	14
	.391	.290	.181	.428	.145	.297	.261	.196	.304	.104
radial loop	1	34	8	4	0	2	27	2	0	0
	.007	.246	.058	.029	.000	.014	.196	.014	.000	.000
arch	5	23	10	2	3	7	22	14	2	4
	.036	.167	.072	.014	.022	.051	.159	.101	.014	.029

TABLE 9 (continued)

	Right Hand					Left Hand				
	1	2	3	4	5	1	2	3	4	5
MILD										
ulnar loop	76	35	84	62	93	82	48	81	68	97
	.628	.239	.694	.512	.769	.678	.397	.669	.562	.802
whorl	42	39	24	56	25	34	30	25	48	22
	.347	.322	.198	.463	.207	.281	.248	.207	.397	.182
radial loop	0	29	4	0	0	1	28	4	1	0
	.000	.240	.033	.000	.000	.008	.231	.033	.008	.000
arch	3	18	9	3	3	4	15	11	4	2
	.025	.149	.074	.025	.025	.033	.124	.090	.033	.017

TABLE 10. Frequency of occurrence of digital patterns among females (730 control, 102 profoundly retarded, 70 severely retarded, 105 moderately retarded, 54 mildly retarded)

	Right Hand					Left Hand				
	1	2	3	4	5	1	2	3	4	5
CONTROL										
ulnar loop	497	290	585	457	643	522	266	506	473	644
	.681	.397	.801	.626	.881	.715	.363	.693	.648	.882
whorl	215	209	93	243	75	179	195	99	223	65
	.295	.286	.127	.333	.103	.245	.267	.136	.305	.089
radial loop	1	122	4	12	2	1	173	31	6	2
	.001	.167	.005	.016	.003	.001	.237	.042	.008	.003
arch	17	109	48	18	10	28	96	94	28	19
	.023	.150	.066	.025	.013	.038	.132	.129	.039	.026
PROFOUND										
ulnar loop	60	30	73	49	84	59	38	69	63	83
	.588	.294	.716	.480	.824	.578	.373	.676	.618	.814
whorl	37	34	19	46	15	33	27	16	33	16
	.363	.333	.186	.450	.147	.324	.265	.157	.324	.157
radial loop	0	20	2	0	0	1	24	2	0	0
	.000	.196	.019	.000	.000	.010	.235	.020	.000	.000
arch	5	18	8	7	3	9	13	15	6	3
	.049	.176	.078	.069	.030	.088	.127	.147	.059	.030

TABLE 10 (continued)

	Right Hand 1	2	3	4	5	Left Hand 1	2	3	4	5
SEVERE										
ulnar loop	39 .557	28 .400	56 .800	36 .514	62 .886	40 .571	19 .271	39 .557	34 .486	50 .714
whorl	28 .400	26 .371	8 .114	29 .414	6 .086	25 .357	26 .371	17 .243	30 .429	15 .214
radial loop	1 .014	8 .114	0 .000	2 .029	0 .000	0 .000	17 .243	4 .057	0 .000	0 .000
arch	2 .029	8 .114	6 .086	3 .043	2 .029	5 .071	8 .114	10 .143	6 .086	5 .071
MODERATE										
ulnar loop	62 .590	35 .333	78 .743	56 .533	76 .724	62 .590	41 .390	81 .771	59 .562	86 .819
whorl	36 .343	41 .390	18 .171	44 .419	25 .238	36 .343	38 .362	15 .143	41 .490	17 .162
radial loop	0 .000	19 .181	1 .009	1 .009	0 .000	0 .000	18 .171	2 .019	1 .009	0 .000
arch	7 .067	10 .010	8 .076	4 .038	4 .038	7 .067	8 .076	7 .067	4 .038	2 .019

TABLE 10 (continued)

	Right Hand					Left Hand				
	1	2	3	4	5	1	2	3	4	5
MILD										
ulnar loop	35	18	42	28	40	39	15	37	29	39
	.648	.333	.778	.519	.740	.722	.278	.685	.537	.722
whorl	16	16	7	25	12	11	16	9	20	12
	.296	.296	.130	.463	.222	.204	.296	.167	.370	.222
radial loop	0	12	2	0	0	2	14	2	1	0
	.000	.222	.037	.000	.000	.037	.259	.037	.019	.000
arch	3	8	3	1	2	2	9	6	4	3
	.056	.148	.056	.019	.037	.037	.167	.111	.074	.056

TABLE 11. Overall frequency of the four digital patterns among control and retarded individuals

MALES

	Control (n=7330)	Profound (n=1350)	Severe (n=1430)	Moderate (n=1380)	Mild (n=1210)	Combined M.R. (n=5370)
Ulnar loop	4350	813	790	852	726	3181
	.593	.602	.553	.616	.600	.592
Whorl	2273	387	448	358	345	1538
	.310	.287	.313	.260	.285	.286
Radial loop	382	55	77	78	67	277
	.052	.041	.054	.057	.055	.052
Arch	325	95	115	92	72	374
	.044	.070	.080	.067	.060	.070

TABLE II (continued)

FEMALES

	Control (n=7330)	Profound (n=1020)	Severe (n=700)	Moderate (n=1050)	Mild (n=540)	Combined M.R. (n=3310)
Ulnar loop	4883	608	403	636	321	1968
	.669	.596	.576	.606	.594	.595
Whorl	1596	276	210	311	144	941
	.219	.271	.300	.296	.267	.284
Radial loop	354	49	32	42	34	157
	.048	.048	.046	.040	.063	.047
Arch	467	87	55	61	41	244
	.064	.085	.079	.058	.076	.073

pattern types accounted for almost 90% of the patterns seen among control individuals

3. radial loops and arches were only rarely seen, and then usually on digit 2 (males - arches=.044, radial loops=.052; females - arches=.064, radial loops=.048).

Several differences were also noted between the two control groups. These included an increased frequency of ulnar loops among the females, with an accompanying decrease in the frequency of whorls.

Generally, the distribution of pattern types among the retarded groups was similar to that seen for the controls. Ulnar loops were the most commonly seen patterns (males=.592; females=.595), whorls were the second most common (males=.286; females=.284), and radial loops and arches were rarely seen except on digit 2 (males - arches= .070, radial loops=.052; females - arches-.073, radial loops=.047). However, several differences were seen between the control and retarded groups, and are listed below.

1. Among the retarded individuals of each sex, there was an overall increase in the frequency of arches (males - .070 vs. .044; females - .073 vs. .064). This was seen in seven of the eight retarded groups, the only exception being moderately retarded females. The increase in arches was particularly noticeable on those digits where arches are rare in controls - digit 1 and digits 4 and 5. For the 1463 control individuals, the frequency of arches on digit 1 was .026; for the 868 retarded individuals, the frequency was .056. Corresponding frequencies

for digit 4 were .022 for the controls and .040 for the retarded individuals; those for digit 5 were .017 for the controls and .033 for the retarded individuals. Increased frequencies of arches among the retarded individuals were also seen on digits 2 and 3, but these were not nearly as striking as those on the three other digits.

2. Although the overall frequency of radial loops was approximately the same for both the control and retarded populations (males - .052 for both groups; females - .048 for the control and .047 for the retarded group), the frequency of radial loops on digit 1 was elevated among the retarded individuals (males - .006 vs. .001; females - .006, vs. .001). Overall frequency of radial loops on digit 1 was .001 for the controls and .006 for the retarded individuals.

3. Among the females only, there was an increase in the frequency of whorls among the retarded individuals. This was accompanied by a decrease in the frequency of ulnar loops in the retarded groups. These findings were constant with all four of the female retarded groups and, for the most part, the shifts in frequencies were seen on all ten digits.

Two 2x4 contingency tables were constructed to determine whether the frequency of the four pattern types were the same for the control and retarded populations. One table was constructed for each sex. Results of the two tests are shown in Table 12; it can be seen that, for each sex, the differences in pattern frequencies were significant

TABLE 12. Chi square test to determine whether there is an equal number of loops, whorls, and arches between the normal and retarded groups

Row 1 = normals
Row 2 = pooled retarded groups (profound-severe and moderate-mild groups combined)

Column 1 = ulnar loop
Column 2 = whorl
Column 3 = radial loop
Column 4 = arch

MALES

Observed -

				total
4350	2273	382	325	7330
3181	1538	277	374	5370
7531	3811	659	699	12700

Expected -

				total
4346.6	2199.6	380.4	403.4	7330
3184.4	1611.4	278.6	295.6	5370
7531	3811	659	699	

Chi square = .003 + 2.449 + .007 + 15.237 + .004 + 3.343 + .009 + 20.793
= 41.845 *

Chi square, $_{.01, 3\ d.f.}$ = 11.345

TABLE 12 (continued)

FEMALES

Observed -

				total
4883	1596	354	467	7300
1968	946	156	240	3310
6851	2542	510	707	10610

Expected -

4713.7	1749.0	350.9	486.4	7300
2137.3	793.0	159.1	220.6	3310
6851	2542	510	707	10610

Chi square = 6.081 + 13.384 + .027 + .774 + 13.411 + 29.420 + .060 + 1.706

= 64.958 *

Chi square$_{.01,\ 3\ d.f.}$ = 11.345

at p=.01. Among the males, the largest factor contributing to the total chi-square score was the difference in the frequency of arches between the control and retarded groups. Among the females, differences in the frequencies of ulnar loops and whorls were the largest contributory factors.

Analysis of the ten-finger patterns

The ten-finger patterns of all individuals were recorded, beginning with digit 1 on the right hand and ending with digit 5 on the left hand.* All ten-finger patterns were then categorized on the basis of the patterns on digits 1, 2 and 3. Initially, each ten-finger pattern was placed into one of five groups, based on the patterns of the two number 1 digits (Table 13). These five groups were ordered primarily on the basis of the various combinations of ulnar loops and whorls on the number 1 digits. Thus, ten-finger patterns with ulnar loops on both number 1 digits were placed into Group 1, those with one ulnar loop and one whorl into either Group 2 or 3, and those with two whorls into Group 4. Due to the relative scarcity of arches and radial loops on digit 1, ten-finger patterns with either of these pattern types on digit 1 were placed into a separate group, Group 5 (Table 14).

* For ease of notation, the initials U (for ulnar loop), W (whorl), R (radial loop), and A (arch) were used in recording the ten-finger patterns. Thus, an individual with seven ulnar loops, whorls bilaterally on digits 4, and a radial loop on digit 2 of the right hand, could be designated as URUWU UUUWU.

TABLE 13. Assignment of three-number codes for all ten-finger patterns falling into one of the first four groups

1st Number		2nd Number		3rd Number	
Digit 1	R L	Digit 2	R L	Digit 3	R L
Group 1 =	U U	1 =	U U	1 =	U U
Group 2 =	U W	2 =	U W	2 =	U W
Group 3 =	W U	3 =	U R	3 =	W U
Group 4 =	W W	4 =	U A	4 =	W W
Group 5 (see Table 14)		5 =	W U	5 =	other
		6 =	W W		
		7 =	W R		
		8 =	W W		
		9 =	R U		
		10 =	R W		
		11 =	R R		
		12 =	R A		
		13 =	A U		
		14 =	A W		
		15 =	A R		
		16 =	A A		

U = ulnar loop W = whorl R = radial loop A = arch

TABLE 14. Assignment of two-number codes for all ten-finger patterns with an arch or radial loop on at least one number 1 digit

1st Number	2nd Number	Digit 1 R	Digit 1 L
5	- =	U	U
	- =	U	W
	3 =	U	R
	4 =	U	A
	- =	W	U
	- =	W	W
	7 =	W	R
	8 =	W	A
	9 =	R	U
	10 =	R	W
	11 =	R	R
	12 =	R	A
	13 =	A	U
	14 =	A	W
	15 =	A	R
	16 =	A	A

U = ulnar loop W = whorl R = radial loop A = arch

Within each of the first four groups, the ten-finger patterns were sub-grouped on the basis of the patterns on digit 2 and digit 3 (Table 13). For example, patterns of digit 2 were divided into 16 categories (reflecting the increased frequency of arches and radial loops on the number 2 digits). Patterns of digit 3 were divided into the same five groups as those for the number 1 digits.

As a result of this grouping, all ten-finger patterns within the first four groups could be represented by a three-number code; such a code specifies six of the ten individual digital patterns. For example, the ten-finger pattern WUUWU WUWUU would be represented as 4-1-2; ten-finger patterns URWWW URRUU and URWRR URRWU would both be represented as 1-11-7.

Because of the relatively small number of ten-finger patterns falling into Group 5, this group was subdivided in a manner different from that used with the other four groups (Table 14). In this group, all ten-finger patterns were coded on the basis of the digit 1 patterns only. The first number for all ten-finger patterns was 5; the second number was assigned depending on the particular types of patterns of digit 1. Thus, ten-finger patterns with an arch on digit 1 of the right hand and a radial loop on digit 1 of the left hand would be given the two number code 5-14; those with an ulnar loop on digit 1 of the right hand and a radial loop on digit 1 of the left hand would be 5-3.

A preliminary analysis was carried out to determine if differences existed between the control and retarded groups with respect to these pattern combinations. For the purpose of this analysis, all ten-finger patterns in the first four groups were placed into 64 different categories based on the first two numbers of their three-number code; thus four of the ten digital patterns could be specified (this analysis did not use all three numbers of the three-number code, due to the fact that this would have generated 320 different cells, most of which would have contained very small numbers). The ten-finger patterns in group 5 were considered as a single category, because of their small number. For the subsequent goodness of fit test, all categories having an expected value greater than one were considered separately (Snedecor and Cochran, 1967). Those categories with expected values less than one were combined with other, similar, categories (e.g., among the males categories 2-4, 2-10, 2-14, and 2-15 were considered together). In instances where no similar category existed, values for cells were arbitrarily put at 0.

Two separate tests were performed, one for males and one for females; so as to maximize the number of observations per cell, the retarded individuals were not subdivided by I.Q. level. Results of the two tests are given in Tables 15 and 16. Among the males, differences between the retarded and control groups were significant at p=.005. Greatest contributions to the total chi-square value (107.87) came from categories 1-4 (4.58), 1-5 (8.13), 2-7 (4.09), and 5 (16.80), all of which involved greater than expected numbers

TABLE 15. Goodness of Fit test to determine whether differences exist between retarded and control males with respect to pattern combinations of digit 1 and digit 2

Row 1 = number of control individuals
Row 2 = number of retarded individuals
Columns = pattern combinations of digit 1 and digit 2

GROUP 1

OBSERVED

1-1	1-2	1-3	1-4	1-5	1-6	1-7	1-8	1-9	1-10	1-11	1-12	1-13	1-14	1-15	1-16
70	13	29	5	17	31	9	1	39	10	43	3	10	1	16	22
66	6	20	11	29	24	5	1	17	11	31	4	11	3	7	22
136	19	49	16	46	55	14	2	56	21	74	7	21	4	23	44

EXPECTED

78.49	10.97	28.28	9.23	26.55	31.74	8.08	1.15	32.32	12.12	42.71	4.04	12.12	2.31	13.27	25.40
57.51	8.03	20.72	6.77	19.45	23.26	5.92	.85	23.68	8.88	31.29	2.96	8.88	1.69	9173	18.60

CHI-SQUARE

.92	.38	.02	1.94	3.44	.02	.10	0	1.38	.37	.00	.27	.37	.74	.56	.46
1.25	.51	.03	2.64	4.69	.02	.14	0	1.88	.51	.00	.37	.51	1.02	.77	.62

TABLE 15 (continued)

GROUP 2

OBSERVED

	2-1	2-2	2-3	2-5	2-6	2-7	2-9	2-11	2-16
	2	1	7	8	11	0	3	6	3
	5	2	1	0	5	3	2	0	0
	7	3	8	8	16	3	5	6	3
EXPECTED									
	4.04	1.73	4.62	4.62	9.23	1.73	2.89	3.46	1.73
	2.96	1.27	3.38	3.38	6.77	1.27	2.11	2.54	1.27
CHI SQUARE									
	1.03	.31	1.23	2.37	.34	1.73	.00	1.86	.93
	1.41	.42	1.67	3.38	.46	2.36	.01	2.54	1.27

TABLE 15 (continued)

GROUP 3

OBSERVED

3-1	3-2	3-3	3-4	3-5	3-6	3-7	3-8	3-9	3-10	3-11	3-12	3-13	3-15	3-16
24	5	6	2	16	39	4	1	7	11	11	4	6	2	10
13	1	4	1	10	13	0	0	6	8	8	2	3	4	1
37	6	10	3	26	52	4	1	13	19	19	6	9	6	11

EXPECTED

21.36	3.46	5.77	1.73	15.01	30.01	2.31	.58	7.50	10.97	10.97	3.46	5.19	3.46	6.35
15.64	2.54	4.23	1.27	10.99	21.99	1.69	.42	5.50	8.03	8.03	2.54	3.81	2.54	4.65

CHI-SQUARE

.33	.67	.01	.04	.07	2.69	1.24	.00	.03	.00	.00	.08	.13	.62	2.10
.45	.91	.01	.06	.09	3.68	1.69	.00	.05	.00	.00	.11	.17	.84	2.87

TABLE 15 (continued)

GROUP 4

OBSERVED

4-1	4-2	4-3	4-4	4-5	4-6	4-7	4-9	4-10	4-11	4-12	4-13	4-16
28	5	3	0	20	95	7	13	15	9	4	0	2
9	4	8	2	17	59	3	6	7	5	0	4	2
37	9	11	2	37	154	10	19	22	14	4	4	4

EXPECTED

21.36	5.19	6.35	1.15	21.36	88.88	5.77	10.96	12.70	8.08	2.31	2.31	2.31
15.64	4.81	4.65	.85	15.64	65.12	4.23	8.04	9.30	5.92	1.69	1.69	1.69

CHI-SQUARE

2.06	.01	1.77	.00	.09	.42	.26	.38	.42	.10	1.24	1.24	1.24
2.82	.01	2.41	.00	.12	.58	.35	.52	.57	.14	1.69	1.69	1.69

TABLE 15 (continued)

GROUP 5

OBSERVED	EXPECTED	CHI-SQUARE
23	39.82	7.10
46	29.18	9.70
69		

TOTAL CHI-SQUARE = 107.87

TABLE 16. Goodness of Fit test to determine whether differences exist between retarded and control females with respect to pattern combinations of digit 1 and digit 2

Row 1 = number of control individuals
Row 2 = number of retarded individuals
Columns = pattern combinations of digit 1 and digit 2

GROUP 1

OBSERVED

1-1	1-2	1-3	1-4	1-5	1-6	1-7	1-8	1-9	1-10	1-11	1-12	1-13	1-14	1-15	1-16
126	21	50	18	23	52	12	1	22	13	40	5	16	1	15	34
34	7	22	5	10	17	5	0	6	3	18	2	6	1	4	7
160	28	72	23	33	69	17	1	28	16	58	7	22	2	19	41

EXPECTED

110.08	10.26	49.54	15.82	22.70	47.47	11.70	.69	19.26	11.01	39.91	4.82	15.14	1.38	13.07	28.21
49.92	8.74	22.46	7.16	10.30	21.53	5.30	.31	8.74	4.99	18.09	2.18	6.86	.62	5.93	12.79

CHI-SQUARE

2.30	.16	.00	.30	.00	.43	.01	0	.39	.36	.00	.01	.05	0	.28	1.19
5.07	.35	.01	.66	.01	.95	.02	0	.86	.79	.00	.02	.11	0	.63	2.62

TABLE 16 (continued)

GROUP 2

	2-1	2-3	2-5	2-6	2-7	2-9	2-11	2-Other
OBSERVED								
	5	3	3	8	0	4	4	6
	10	2	1	8	5	1	2	4
	15	5	4	16	5	5	6	10
EXPECTED								
	10.32	3.44	2.75	11.01	3.44	3.44	4.13	6.88
	4.68	1.56	1.25	4.99	1.56	1.56	1.87	3.12
CHI-SQUARE								
	2.74	.06	.02	.82	3.44	.09	.00	.11
	6.05	.12	.05	1.82	7.59	.20	.01	.25

TABLE 16 (continued)

GROUP 3

OBSERVED

	3-1	3-2	3-3	3-5	3-6	3-7	3-9	3-10	3-11	3-16	3-Other
	14	3	5	9	16	3	2	4	4	2	6
	8	3	0	6	16	4	2	3	4	2	0
	22	6	5	15	32	7	4	7	8	4	6
EXPECTED											
	15.14	4.13	3.44	10.32	22.02	4.82	2.75	4,82	5.50	2.75	4.13
	6.86	1.87	1.56	4.68	9.98	2.18	1.25	2.18	2.50	1.25	1.87
CHI-SQUARE											
	.09	.31	.71	.17	1.65	.69	.20	.14	.41	.20	.85
	.19	.68	1.56	.37	3.63	1.52	.45	.31	.90	.45	1.87

TABLE 16 (continued)

GROUP 4

	4-1	4-2	4-3	4-5	4-6	4-7	4-9	4-10	4-11	4-13	4-15	4-16	4-Other
OBSERVED													
	20	8	5	7	64	9	4	3	11	3	4	3	4
	8	5	0	5	37	1	3	3	5	1	0	2	1
	28	13	5	12	101	10	7	6	16	4	4	5	5
EXPECTED													
	19.26	8.94	3.44	8.26	69.49	6.88	4.82	4.13	11.01	2.75	2.75	3.44	3.44
	8.74	4.06	1.56	3.74	31.51	3.12	2.18	1.87	4.99	1.25	1.25	1.56	1.56
CHI-SQUARE													
	.03	.10	.71	.19	.43	.65	.14	.31	.00	.02	.57	.06	.09
	.06	.22	1.56	.42	.96	1.44	.31	.68	.00	.05	1.25	.12	.20

TABLE 16 (continued)

GROUP 5

OBSERVED	EXPECTED	CHI-SQUARE
35	46.10	2.67
32	20.90	5.90
67		

TOTAL CHI-SQUARE = 76.99

of retarded individuals, and from 2-5 (5.85), 2-11 (4.40), 3-6 (6.37), 3-16 (4.97), and 401 (4.88), involving an excess of control individuals. Together these nine categories accounted for over one-half of the total chi-square value (59.97/107.87). Among the females, the differences were significant at p=.01. Categories in which large values were seen included 2-1 (8.79), 2-7 (10.03), 3-5 (5.28), and 5 (8.57), involving an excess in the retarded group, and 1-1 (7.37), and 1-5 (8.13), in which a greater than expected number of control individuals were seen. Together, these categories accounted for over one-half of the total chi-square value seen in the females (43.85/76.99).

Further analysis of the ten-finger patterns was aimed at the detection of ten-finger patterns seen in excess among the retarded groups. Because the examination of total finger ridge count, pattern intensity, and frequency distribution of the four pattern types had shown no significant differences between the various levels of retardation, for all further tests the retarded individuals were divided into only two groups: profound-severe retardation and moderate-mild retardation.

The initial analysis of pattern combinations (Tables 15 and 16) had revealed an excess of retarded males among categories in groups 1 and 5; among retarded females, excesses were seen in groups 2, 3, and 5. Thus, it was decided to further examine these groups to determine if they contained any ten-finger patterns particularly likely to be associated with mental retardation. Because our earlier analysis of pattern distribution had shown arches to be increased in frequency on all digits in the retarded population, it was decided to extend the analysis of group 5 to include an analysis of arches in general. Thus, the following groups of ten-finger patterns were evaluated to determine if any contained groups of retarded individuals with similar ten-finger patterns not typically seen among the control individuals:

1. individuals with arches on any digit
2. individuals with radial loops on either number I digit (from group 5)
3. females with an ulnar loop on one number I digit and a whorl on the other number I digit (groups 2 and 3)
4. males with ulnar loops bilaterally on the number I digits (group I).

A summary of the statistical findings with respect to these groups is given in Table 32.

Analysis of pattern types seen to be elevated in the retarded population

Analysis of individuals with arches on any finger

Based on the results of the earlier tests involving pattern distribution and pattern combinations, this general heading was divided into four categories: overall number of arches among individuals with at least one arch; analysis of arches on digit I; analysis of arches on digit 4; analysis of arches on digit 5. For all four categories, the goal was to detect groups of ten-finger patterns with high mentally retarded:control ratios (i.e., ten-finger patterns apparently associated with mental retardation).

Overall number of arches in individuals with at least one arch

A total of 342 control individuals (148 males and 194 females) and 227 retarded individuals (145 males and 82 females) had at least one arch. The distribution of the number of arches among these individuals is shown in Table 17.

The frequency of individuals with at least one arch was only slightly higher among the retarded individuals than the control individuals (retarded:control=1.12:1.00). In fact, among the females the frequency of individuals with at least one arch was actually higher among the control group. However, if all individuals with at least one arch were subdivided into two groups, those with a low number of arches (105), and those with a high number (6-10), differences between the control and retarded groups became apparent (Table 18). Among those individuals with 1-5 arches, the overall retarded:control ratio was almost 1:1 (slightly higher among the males and slightly lower among the females). However, among those individuals with arches on more than half of the ten digits, there was an increased frequency among the retarded individuals when compared to the controls (overall retarded:control=1.82:1.00). This increase was more noticeable in the males than the females, and was seen in both I.Q. levels within each sex.

TABLE 17. Number of arches in individuals with at least one arch

n= # of arches	MALES 733 Control	278 Profound-Severe	259 Moderate-Mild	FEMALES 730 Control	172 Profound-Severe	159 Moderate-Mild
1	70	25	32	92	17	14
2	39	20	17	35	9	7
3	18	10	9	18	7	2
4	8	5	3	19	4	3
5	5	5	5	13	3	3
6	4	3	0	11	5	1
7	1	2	0	0	0	1
8	0	1	1	4	1	3
9	0	2	2	2	0	0
10	3	2	1	0	1	1
n=	148	75	70	194	47	35
frequency=	.202	.270	.270	.266	.273	.220
M.R.:control		1.34:1	1.34:1		1.03:1	.83:1
M.R.:control		1.34:1			.93:1	
(combined by sex)				1.12:1		

TABLE 18. Individuals with 1-5 arches and with 6-10 arches

number of individuals with 1-5 arches:

n=	140	65	66	177	40	29
frequency=	.191	.234	.255	.242	.233	.182
M.R.:control		1.22:1	1.33:1		.96:1	.75:1
M.R. control (combined by sex)		1.28:1			.86:1	
total M.R.:control				1.06:1		

number of individuals with 6-10 arches

n=	8	10	4	17	7	6
frequency=	.011	.036	.015	.023	.041	.038
M.R.:control		3.30:1	1.42:1		1.75:1	1.62:1
M.R.:control (combined by sex)		2.36:1			1.70:1	
Total M.R.:control				1.82;1		

Analysis of arches on digit I

Table 19 shows the number of control and retarded individuals with arches on one or both number I digits. The frequency of individuals with an arch on one or both number I digits was found to be higher among the retarded individuals than the control individuals. Altogether, the frequency of individuals with an arch on one or both number I digits was 0.038 (55/1463) among the controls and 0.079 (69/868) among the retarded individuals. This translates to a 2.12:1.00 retarded to control ratio. The difference in frequencies between the control and retarded individuals was more striking among the males than the females and among the profound-severe group than the moderate-mild group.

Analysis of individuals with an arch on one or both number I digits and 6-10 arches overall revealed an even higher retarded: control ratio (Table 20). Again, the differences seen between the control and retarded groups were more apparent among the males, and among the profound-severe groups.

Among normal individuals, arches on digit I are commonly seen in association with arches or radial loops on digit 2 (Slatis, personal communication). Thus, in an attempt to further improve upon the retarded:control ratio within the arch on digit I group, all individuals with an arch on at least one number I digit and no arch or radial loop on either digit 2 were examined. Findings are summarized in Table 21. Although the number of individuals is small, it can be seen that the retarded individuals were much more likely

TABLE 19. Frequency of control and retarded individuals with arches on one or both number I digits

	MALES			FEMALES		
overall n=	(733) Control	(278) Profound Severe	(259) Moderate Mild	(730) Control	(172) Profound Severe	(159) Moderate Mild
Arch on right digit I	1	5	2	5	2	3
Arch on left digit I	11	13	5	16	9	6
Arches on both digits I	10	10	6	12	5	3
Total	22	28	13	33	15	12
Frequency	.030	.101	.050	.045	.093	.076
M.R.:control		3.36:1	1.67:1		2.06:1	1.76:1
Combined M.R.:control (by sex)		2.53:1			1.89:1	
Total M.R.:control				2.12:1		

TABLE 20. Individuals with an arch on one of both number I digits and 6-10 arches overall

	MALES			FEMALES		
overall n=	(733) Control	(278) Profound Severe	(259) Moderate Mild	(730) Control	(172) Profound Severe	(159) Moderate Mild
n	5	8	4	7	5	3
Frequency	.007	.029	.015	.010	.029	.019
M.R.:control		4.22:1	2.26:1		3.03:1	1.97:1
Combined M.R.:control		3.14:1			2.40:1	
Total M.R.:control				2.81:1		

TABLE 21. Number of individuals with an arch on at least one number 1 digit and no arch or radial loop on either number 2 digit

	MALES			FEMALES		
overall n=	(733)	(278)	(259)	(730)	(172)	(159)
	Control	Profound Severe	Moderate Mild	Control	Profound Severe	Moderate Mild
Arch on right digit 1	0	1	1	0	0	0
Arch on left digit 1	1	5	1	0	3	5
Arches on both digits 1	3	1	0	1	0	0
Total	4	7	2	1	3	5
Frequency	.005	.025	.007	.001	.017	.031
M.R.:Control		4.61:1	1.42:1		12.73:1	22.96:1
M.R.:Control (combined by sex)		3.07:1			17.64:1	
Total M.R.:Control				5.73:1		

TABLE 22. Individuals with an arch on at least one number 1 digit and a whorl(s) on any finger

	MALES			FEMALES		
overall n=						
	Control	Profound Severe	Moderate Mild	Control	Profound Severe	Moderate Mild
Arch on right digit 1	0	4	1	1	0	1
Arch on left digit 2	7	5	3	3	4	5
Arch on both digits 1	2	1	3	2	2	0
Total	9	10	7	6	6	6
Frequency	.012	.036	.027	.008	.035	.038
M.R.:Control			2.03:1	2.20:1	4.24:1	4.59:1
M.R.:Control (combined by sex)			2.58:1		4.41:1	
Total M.R.:Control				3.26:1		

to have this configuration than were the normals (overall retarded: control ratio = 5.73:1.00). Furthermore, when individuals with arches on both number 1 digits were removed, the difference between the retarded and control groups became even more pronounced. Only one control and 16 retarded individuals had this particular type of ten-finger pattern, yielding a retarded:control ratio of 26.96:1.00.

Arches and whorls are generally not seen together in the same ten-finger pattern (Poll, 1931). Thus, another attempt to improve upon the retarded:control ratio involved examining all individuals with an arch on at least one number 1 digit, and a whorl on any digit. Results are shown in Table 22. Again, the numbers involved were very small, but indicated an increase among the retarded groups for this particular type of ten-finger pattern (retarded:control ratio = 3.26:1.00).

Analysis of arches on digit 4

The number of control and retarded individuals with at least one arch on digit 4 is shown in Table 23. The proportion of individuals with an arch on digit 4 was higher among the retarded than the control population (overall retarded:control ratio = 2.15:1.00). However, these data were confounded by the fact that many of the individuals within this group also had arches on digit 1, a configuration already known to involve an excess of retarded individuals. Evaluation of the data, minus those individuals, is also shown in Table 23. An excess of retarded individuals can still

TABLE 23. Control and retarded individuals with at least one arch on digit 4

	MALES			FEMALES		
overall n=	(733)	(278)	(259)	(733)	(172)	(159)
	Control	Profound Severe	Moderate Mild	Control	Profound Severe	Moderate Mild
n	18	16	8	27	15	12
frequency	.018	.058	.031	.037	.087	.075
M.R.:Control		3.25:1	1.74:1		2.36:1	2.04:1
M.R.:Control (combined by sex)		2.52:1			2.21:1	
Total M.R.:Control				2.15:1		

Minus individuals with an arch on at least one number 1 digit

n	9	6	4	21	11	8
frequency	.012	.022	.015	.029	.064	.050
M.R.:Control		1.76:1	1.26:1		2.22:1	1.75:1
M.R.:Control (combined by sex)		1.52:1			1.99:1	
Total M.R.:Control				1.63:1		

be seen, although not nearly so pronounced as previously (1.63:1.00).

Analysis of arches on digit 5

A total of 32 control and 41 retarded individuals had an arch on at least one number 5 digit (Table 24). This represents a 2.16:1.00 retarded:control ratio. However, many of these individuals had arches on either digit 1 or digit 4, configurations already known to be increased among the retarded population. Examination of the data, minus those individuals with arches on either digit 1 or digit 4, reduced the retarded:control ratio to 1.26:1.00 (Table 24).

Analysis of individuals with a radial loop on at least one number 1 digit

The number of control and retarded individuals with a radial loop on one or both number 1 digits is shown in Table 25. The occurrence of this pattern on digit 1 was rare among both the control and retarded groups; overall frequency among the control group was .002 (3/1463), while that of the retarded individuals was .010 (9/868). The overall retarded:control ratio was 5.06:1.00.

Analysis of females with an ulnar loop on one number 1 digit and a whorl on the other number 1 digit (groups 2 and 3)

Examination of females with ten-finger patterns falling into group 2 or group 3 revealed a greater proportion of retarded individuals than controls for both groups (Table 26). The retarded:

TABLE 24. Control and retarded individuals with an arch on at least one number 5 digit

	MALES			FEMALES		
overall n=	(733)	(278)	(259)	(730)	(172)	(159)
	Control	Profound Severe	Moderate Mild	Control	Profound Severe	Moderate Mild
n	13	14	9	19	9	9
Frequency	.018	.050	.035	.026	.052	.057
M.R.:Control		3.71:1	1.96:1		2.01:1	2.17:1
M.R.:Control (combined by sex)		2.41:1			2.09:1	
Total M.R.:Control				2.16:1		

Minus individuals with an arch on digit 1 or digit 4

	MALES			FEMALES		
	Control	Profound Severe	Moderate Mild	Control	Profound Severe	Moderate Mild
n	5	3	2	7	2	2
Frequency	.007	.011	.007	.010	.012	.013
M.R.:Control		1.58:1	1.13:1		1.21:1	1.31:1
M.R.:Control (combined by sex)		1.35:1			1.26:1	
Total M.R.:Control				1.26:1		

TABLE 25. Control and retarded individuals with a radial loop on at least one number I digit

	MALES			FEMALES		
overall n=	(733)	(278)	(259)	(730)	(172)	(159)
	Control	Profound Severe	Moderate Mild	Control	Profound Severe	Moderate Mild
Radial loop on digit I on right hand	0	2	0	I	I	0
Radial loop on digit I on left hand	I	0	2	I	I	2
Radial loops on both number I digits	0	0	I	0	0	0
Total	I	2	3	2	2	2
Frequency	.001	.007	.012	.003	.012	.013
M.R.:Control		5.27:I	8.49:I		4.24:I	4.59:I
M.R.:Control (combined by sex)		6.83:I			4.41:I	
Total M.R.:Control				5.06:I		

TABLE 26. Number of female control individuals with an ulnar loop on one number I digit and a whorl on the other number I digit

	Group 2 (u---- w----)			Group 3 (w---- u----)		
overall n=	(730)	(172)	(159)	(730)	(172)	(159)
	Control	Profound Severe	Moderate Mild	Control	Profound Severe	Moderate Mild
n	33	18	15	68	25	23
Frequency	.045	.105	.094	.093	.145	.145
M.R.:Control		2.31:1	2.09:1		1.56:1	1.56:1
M.R.:Control (combined by sex)		2.31:1			1.56:1	
Total M.R.:Control				1.77:1.00		

control ratio was higher for those ten-finger patterns in group 2 (2.21:1.00) than for those in group 3 (1.56:1.00). Subsequent analysis of groups 2 and 3 was aimed at maximizing the retarded:control ratio, and included separate analyses of patterns on digit 2, digit 3, digit 4 and digit 5.

Analysis of digit 2

Digit 2 patterns of the 101 control and 81 retarded females with ten-finger patterns falling into groups 2 or 3 are listed in Table 27. From the analysis of pattern combinations (Table 16), it was already known that, for groups 2 and 3, a greater than expected number of retarded females had ulnar loops on both number 2 digits (19 controls vs. 18 retarded females; retarded:control ratio = 2.09:1.00). Additionally, the combination radial loop-whorl or whorl-radial loop on the right and left number 2 digit was found in .011 (8/733) control females and in .039 (.3/331) of the retarded females (retarded:control = 3.58:1.00).

Analysis of digit 3

Patterns on digit 3 of all females with ten-finger patterns in group 2 or 3 are shown in Table 28. Because analysis revealed no difference between control and retarded individuals with respect to occurrence of radial loops or arches on digit 3, these patterns were grouped together into one category (i.e., other). Analysis of digit 3 patterns revealed an excess of retarded females with

TABLE 27. Distribution of patterns of digit 2 among individuals with ten-finger patterns falling into groups 2 or 3

Digit 2		Group 2 (u---- w----)			Group 3 (w---- u----)		
Right	Left	Control	Profound Severe	Moderate Mild	Control	Profound Severe	Moderate Mild
U	U	5	3	7	14	6	2
U	W	0	0	0	3	2	1
U	R	3	1	1	5	0	0
U	A	3	0	0	1	0	0
W	U	3	1	0	9	1	5
W	W	8	6	2	16	6	10
W	R	0	4	1	3	3	1
W	A	0	0	0	0	0	0
R	U	4	1	0	2	2	0
R	W	1	0	1	4	1	2
R	R	4	0	2	4	2	2
R	A	0	0	0	0	0	0
A	U	0	1	1	3	0	0
A	W	0	0	0	0	0	0
A	R	0	0	0	2	0	0
A	A	2	1	0	2	2	0
n=		33	18	15	68	25	23
overall n=		730	172	159	730	172	159

TABLE 28. Patterns of digit 3 among females with ten-finger patterns falling into Group 2 or Group 3

Patterns of digit 3		GROUP 2			GROUP 3		
Right	Left	Control	Profound Severe	Moderate Mild	Control	Profound Severe	Moderate Mild
U	U	23	9	11	40	16	12
U	W	1	4	1	4	4	2
W	U	0	1	0	6	0	4
W	W	3	2	1	9	1	4
OTHER		6	2	2	9	4	1
		33	18	15	68	25	23

W-U or U-W on the number 3 digits

	Control	Profound Severe	Moderate Mild	Control	Profound Severe	Moderate Mild
overall n=	(730)	(172)	(159)	(730)	(172)	(159)
n=	1	5	1	10	4	6
Frequency	.001	.029	.006	.014	.023	.038
M.R.:Control		21.22:1	4.59:1		1.70:1	2.75:1
M.R.:Control (combined by group)		13.23:1			2.21:1	
Total M.R.:Control				3.20:1		

the combination whorl-ulnar loop or ulnar loop-whorl on digit 3. This was more procounced for Group 2 (retarded:control - 13.23:1.00) than group 3 (2.21:1.00). However, the increase was consistent for both groups and for both I.Q. levels within each group. The total retarded:control ratio was 3.20:1.00, almost twice the ratio for groups 2 and 3 in general.

Analysis of digit 4

Table 29 shows the patterns of digit 4 of females with ten-finger patterns in groups 2 and 3. Among the control females of both groups 2 and 3, the majority of the ten-finger patterns included ulnar loops bilaterally on digit 4. This was not the case with the retarded females, resulting in an increased retarded:control ratio for ten-finger patterns falling into groups 2 or 3 but without the combination ulnar loop-ulnar loop on digit 4. The increase in the retarded:control ratio was more noticeable among the retarded females of group 2 than those of group 3.

Analysis of digit 5

Analysis of digit 5 patterns among females with ten-finger patterns in groups 2 or 3 revealed an excess of retarded females with a whorl on one or both number 5 digits (Table 30). The retarded: control ratio (3.67:1.00) was approximately twice as great as the overall ratio for groups 2 and 3 together. Again, the increase in the frequency of retarded females with this pattern was more marked

TABLE 29. Patterns of digit 4 among females with ten-finger patterns in Groups 2 and 3

Patterns of digit 4 Right	Left	GROUP 2 Control	Profound Severe	Moderate Mild	GROUP 3 Control	Profound Severe	Moderate Mild
U	U	21	6	6	34	9	7
U	W	3	1	1	2	4	1
W	U	2	1	3	11	3	7
W	W	6	9	3	20	8	8
OTHER		1	1	2	1	1	0
		33	18	15	68	25	23

Not U-U on digit 4

	GROUP 2 Control	Profound Severe	Moderate Mild	GROUP 3 Control	Profound Severe	Moderate Mild
n	12	12	9	34	16	16
Frequency	.016	.070	.056	.047	.093	.094
M.R.:Control		4.24:1	3.44:1		2.00:1	2.16:1
M.R.:Control (combined by group)		3.86:1			2.08:1	
Total M.R.:Control				2.54:1		

TABLE 30. Patterns of digit 5 among females with ten-finger patterns in groups 2 or 3

Patterns of digit 5 Right	Left	GROUP 2 Control	GROUP 2 Profound Severe	GROUP 2 Moderate Mild	GROUP 3 Control	GROUP 3 Profound Severe	GROUP 3 Moderate Mild
U	U	28	10	13	56	19	13
U	W	3	6	0	3	2	0
W	U	0	0	1	4	3	5
W	W	1	2	1	4	0	5
OTHER		1	0	0	1	1	0
		33	18	15	68	25	23

W on either or both number 5 digits

Patterns of digit 5 Right	Left	GROUP 2 Control	GROUP 2 Profound Severe	GROUP 2 Moderate Mild	GROUP 3 Control	GROUP 3 Profound Severe	GROUP 3 Moderate Mild
U	W	3	6	0	3	2	0
W	U	0	0	1	4	3	5
W	W	1	2	1	4	0	5
n=		4	8	2	11	5	10
Frequency		.005	.047	.013	.015	.035	.063
M.R.:Control			8.39:1	2.30:1		1.93:1	4.17:1
M.R.:Control (combined by group)			5.51:1			3.01:1	
Total M.R.:Control					3.67:1		

among individuals in group 2 than those in group 3.

Analysis of males with ulnar loops on both number 1 digits (group 1)

A total of 319 control males and 268 retarded males were seen to have ulnar loops on both number 1 digits (Table 31). This yielded a slightly higher than expected retarded:control ratio (1.15:1). Within this general heading, three types of ten-finger patterns were observed with substantially higher retarded:control ratios:

1. Males with a whorl on digit 2 of the right hand and an ulnar loop on digit 2 of the left hand; 29 retarded and 17 control individuals had this particular pattern, giving a M.R.:control ratio of 2.33:1.
2. Males with an arch on one number 2 digit and either an ulnar loop or a whorl on the other number 2 digit (U-A or A-U or W-A or A-W); the combined retarded:control ratio for these configurations was 2.09:1.
3. Males with a whorl on one or both number 5 digits; the retarded: control ratio for this pattern was 1.49:1; however when all individuals with the sequence ---WU on one hand (i.e., a whorl on digit 4 and an ulnar loop on digit 5) were removed from this group, the M.R.:control ratio increased to almost 2:1 (1.99:1).

TABLE 31. Ten-finger patterns of males with ulnar loops on both number 1 digits

	Control	Profound Severe	Moderate Mild
Overall			
n	319	132	136
frequency	.435	.474	.525
M.R.:control		1.09:1	1.21:1
Total M.R.:control		1.15:1	
W-U on digit 2			
n	17	15	14
frequency	.023	.054	.054
M.R.:control		2.02:1	2.16:1
Total M.R.:control		2.09:1	
A-U or U-A or W-A or A-W on digit 2			
n	17	13	13
frequency	.023	.047	.050
M.R.:control		2.02:1	2.16:1
Total M.R.:control		2.09:1	
W on either number 5 digit			
n	42	29	17
frequency	.057	.104	.066
M.R.:control		1.82:1	1.15:1
Total M.R.:control		1.49:1	
W on either number 5 digit, not ---WU on either hand			
n	22	22	10
frequency	.030	.079	.039
M.R.:control		2.64:1	1.29:1
Total M.R.:control		1.99:1	

Summary of the analysis of ten-finger patterns

Table 32 shows the retarded:control ratios for seven general categories of ten-finger patterns seen in excess among mentally retarded individuals of one or both sexes. Within three of these categories, ten-finger patterns were further analyzed to yield additional types of ten-finger patterns found in excess among the retarded individuals.

Chi-square values were assigned to 18 groups of ten-finger patterns listed in Table 32. These values were the result of 18 different 2x2 contingency tables, in which the number of control and retarded individuals with the pattern in question were compared to the number of individuals without the pattern. For example, for individuals with a radial loop on digit I, the following table was constructed:

	Radial loop digit I	Patterns other than radial loop on digit I	Total
Control	3	1460	1463
Retarded	9	859	868
	12	2219	2331

Chi square = 7.36

The chi-square values cannot be accurately evaluated as statistical evidence, because in many cases they resulted from posterior searches for particular types of ten-finger patterns. However, many of the chi-square values were large enough that it is clear that evidence

TABLE 32. Types of ten-finger patterns found in excess among mentally retarded individuals

	M.R.	Control	M.R.: Control	Chi-Square
I. females with a whorl on one number 1 digit and an ulnar loop on the other number 1 digit	81	101	1.77:1	18.11
A. and without ulnar loops bilaterally on the number 4 digits	53	46	2.54:1	25.38
B. and a whorl on one or both number 5 digits	25	15	3.67:1	18.98
C. and a whorl on one number 3 digit and an ulnar loop on the other number 3 digit	16	11	3.20:1	10.16
D. and the sequence whorl-radial loop or radial loop-whorl on the number 3 digits	13	19	2.09:1	9.41
E. and ulnar loops on both number 2 digits	18	19	2.09:1	5.43
II. individuals with an arch on at least one number 1 digit	68	55	2.12:1	18.10
A. and without an arch or radial loop on either number 2 digit	17	5	5.73:1	27.80
B. and a whorl(s) on any digit	29	15	3.26:1	15.77
C. and 6-10 arches	20	12	2.81:1	8.86
III. individuals with an arch on at least one number 4 digit	51	40	2.15:1	14.33
IV. individuals with an arch on at least one number 5 digit	41	32	2.16:1	11.55
V. individuals with a radial loop on one or both number 1 digits	9	3	5.06:1	7.36
VI. males with ulnar loops on both number 1 digits	268	319	1.15:1	5.08
A. and the sequence whorl-ulnar loop on the right and left number 2 digits	29	17	2.33:1	8.43
B. and a whorl on either number 5 digit but without the sequence ---WU on the other hand	32	22	1.99:1	6.65

TABLE 32 (continued)

	C. and an arch on one number 2 digit and whorl or ulnar loop on the other number 2 digit	26	17	2.09:1	6.03
VII.	individuals with 6-10 arches	27	25	1.82:1	4.91

supports a conclusion of difference between control and retarded individuals. The chi-square values act as rough indicators of the various M.R.:control ratios, by taking into account the number of individuals found with the particular type of ten-finger pattern under study. For example, the chi-square value for arches on digit I is greater than that for radial loops on digit I, even though the M.R.:control ratio is greater for radial loops on digit I. This reflects the fact that many more individuals were seen with arches on digit I than with radial loops on digit I.

A comparison of I.Q. levels among retarded groups with respect to the different types of pattern combinations in excess among retarded individuals is summarized in Table 33. This table lists the seven general categories in Table 32 again, this time with respect to the retarded:control ratios for the four retarded groups. From the table it can be seen that the ratios are little different between the profound-severe and moderate-mild groups, for all ten-finger patterns except those involving arches. However, among those types of ten-finger patterns only, there is a consistently elevated ratio among the profoundly or severely retarded males when compared to the other categories (i.e., moderately or mildly retarded males and both groups of retarded males).

Selection of retarded individuals to be further examined

Subsequent examination of the 18 types of ten-finger patterns in Table 32 was aimed at the selection of at least one study (i.e.,

TABLE 33. Retarded:control ratios among different levels of retardation with respect to seven categories of ten-finger patterns seen in excess among retarded individuals

		Profound-Severe	Moderate-Mild	Profound-Severe	Moderate Mild
I.	Females with a whorl on one number 1 digit and an ulnar loop on the other	-	-	1.81:1	1.72:1
II.	Individuals with an arch on at least one number 1 digit	3.36:1	1.67:1	2.06:1	1.67:1
III.	Individuals with an arch on at least one number 4 digit	3.25:1	1.74:1	2.36:1	2.04:1
IV.	Individuals with an arch on at least one number 5 digit	3.71:1	1.96:1	2.01:1	2.17:1
V.	Individuals with a radial loop on at least one number 1 digit	5.27:1	8.59:1	4.24:1	4.59:1
VI.	Males with ulnar loops on both number 1 digits	1.09:1	1.21:1	-	-
VII.	Individuals with 6-10 arches	3.30:1	1.42:1	1.75:1	1.62:1

with respect to physical examination, family history, and possible biochemical or chromosomal abnormalities). Two criteria were used in this selection process:

1. an obvious excess of retarded individuals in the group (i.e., a retarded:control ratio of at least 2:1)
2. a relatively large number of retarded individuals in the group (i.e., at least 5% of the total retarded sample, or 44 individuals).

Three of the groups listed in Table 32 fit these criteria; individuals with an arch on at least one number 1 digit, and females with an ulnar loop on one number 1 digit and a whorl on the other number 1 digit, and females with an ulnar loop on one number 1 digit and a whorl on the other but without ulnar loops bilaterally on the number 4 digits.

Because of several reports which linked the occurrence of excess arches and radial loops with various mental retardation disorders (Lewandowski and Yunis, 1975; Preus and Fraser, 1972), it was decided that all individuals with an arch on at least one number 1 digit would be further analyzed. Subsequently, individuals with a radial loop on at least one number 1 digit were added to this group. Hence, the eventual group of retarded individuals selected for further evaluation included all individuals with non-normal thumbprint patterns (i.e., arches or radial loops).

Examination of retarded individuals with an arch or radial loop on at least one number I digit

Forty-seven retarded individuals with an arch or radial loop on at least one number digit were available for examination; 30 other retarded individuals with an arch or radial loop on digit I had been either released from institutional care or had died. The 47 individuals consisted of 28 males (12 profoundly retarded, 12 severely retarded, three moderately retarded, and one mildly retarded) and 19 females (ten profoundly retarded, six severely retarded, and three moderately retarded). Subsequent examination included a physical evaluation of all individuals, review of all their records (with respect to previous physical examinations, family and developmental histories, and results of urine and blood screens for metabolic errors), and cytogenetic analysis. A summary of the findings is shown in Table 34.

Physical examination

Twenty-one of the 47 individuals were found to have multiple physical anomalies (i.e., three or more). Of the remaining 26, 13 had two abnormalities and six individuals had one abnormality. Seven individuals appeared to be free of any physical stigmata. The most commonly seen abnormalities included high arched palate (18/47), malformed or low-set ears (8/47), strabismus (8/47), broad nose (8.47), cryptorchidism (8/28 males; four bilateral and three unilateral), endophthalmus (7/47), seizures (6/47), and scoliosis

TABLE 34. Summary of findings among 47 mentally retarded individuals with an arch or radial loop on at least one number I digit

Case no.	Ten-finger pattern	Sex	Age	Level of Retardation	Mat. age at birth	Pat. age at birth
1.	RAAUU UAAUU	M	17	severe	38	38
2.	UAUUU AAUUU	F	48	severe	34	34
3.	UAAUU AAAUU	M	20	profound	35	34
4.	UAAAU AAAAU	M	25	severe	33	35
5.	RWWWW WWWWW	M	20	severe	25	26
6.	AUUWU AUUUU	M	25	severe	35	44
7.	UAAWU AAAUU	F	38	profound	25	22
8.	AUAUU AAUAU	M	16	profound	31	42
9.	AAAAA AAAAA	M	13	severe	23	38
10.	AUUAU ARAAA	M	24	profound	25	32
11.	WAAAU RAAAU	F	22	profound	22	26
12.	ARUUU URUUU	M	56	moderate	18	28
13.	RAUWA WAAWU	F	29	severe	19	21
14.	UUUUU AUAUU	F	67	profound	22	26
15.	AAAAU AAAAA	F	31	profound	28	33
16.	AUAUU AAAUU	F	17	profound	23	30
17.	UUUUU ARAUU	M	17	profound	unknown	unknown
18.	AAUUU AAAUU	M	16	profound	36	32
19.	AAUUA UUUUU	F	31	profound	unknown	unknown
20.	UWWWW ARWWW	M	15	profound	22	22
21.	AUAWU UAUWU	M	15	profound	27	34
22.	UUAUU AUAAU	F	26	severe	27	32
23.	URWWU RWWWU	M	26	moderate	unknown	unknown
24.	UWUWU AUUUU	M	22	profound	27	33
25.	AAAWU UAUUU	M	31	profound	35	43
26.	UAUWU AAUWW	M	37	mild	20	31
27.	URUWU ARUUU	F	18	profound	34	36
28.	WAUWU AAUAU	M	49	severe	28	44
29.	URRUU ARUUU	M	35	severe	38	37
30.	AAAAA AAAAA	M	34	severe	37	29
31.	UUUUU AUUUU	F	20	profound	34	44
32.	ARUWW AAUWU	F	17	moderate	17	22
33.	ARWUU ARWUA	F	22	profound	30	28
34.	UUUUU AAUUA	M	15	moderate	22	22
35.	UAUWU AUUWW	F	6	profound	20	22
36.	AAUWU UUUWU	M	54	severe	18	22
37.	UUUUU AUWWW	M	39	severe	27	32
38.	UWWWU AWUUU	M	11	severe	25	25
39.	AAUUU AAUAA	M	21	profound	31	33

TABLE 34 (continued)

Case no.	Ten-finger pattern	Sex	Age	Level of Retardation	Mat. age at birth	Pat. age at birth
40.	AAAAA AAAAA	F	47	moderate	25	29
41.	UUUWW AUUWW	F	25	moderate	22	29
42.	AAAAA AAAAA	F	17	severe	27	29
43.	ARAUU UAAUU	M	20	severe	26	31
44.	AAAUU AAAWW	F	58	severe	26	28
45.	UUUUU AAAUU	M	10	profound	25	26
46.	UAUUU AAUUU	M	18	profound	unknown	unknown
47.	UUAUU AAAAA	F	33	severe	23	24

TABLE 34 (continued)

Case no.	Birthweight (lbs. - oz.)	Chromosome analysis	Biochemical abnormality	Family history
1.	7 - 14	47,XY +dic.	-	-
2.	6 - 8	47,XX +dic.	-	-
3.	7 - 2	46,XY (15q+)	-	-
4.	7 - 14	47,XX +mar.	-	-
5.	6 - 3	46,XY	-	-
6.	2 - 1	46,XY	-	-
7.	6 - 13	46,XX	-	-
8.	unknown	46,XY	↑ phe.	+
9.	6 - 3	46,XY	↑ muco.	+
10.	unknown	46,XY	-	-
11.	8 - 3	46,XX	-	-
12.	9 - 8	46,XY	-	-
13.	6 - 12	46,XX	-	-
14.	4 - 8	46,XX	-	-
15.	5 - 0	46,XX	-	-
16.	6 - 12	46,XX	-	-
17.	8 - 4	46,XY	-	unknown
18.	6 - 10	46,XY	-	-
19.	6 - 2	46,XX	-	unknown
20.	6 - 8	46,XY	-	-
21.	8 - 4	46,XY	-	-
22.	6 - 0	46,XX	-	-
23.	7 - 0	46,XY	-	-
24.	6 - 0	46,XY	-	-
25.	9 - 8	46,XY	-	-
26.	8 - 4	46,XY	-	-
27.	7 - 3	46,XX	-	-
28.	8 - 0	46,XY	-	+
29.	6 - 8	46,XY	-	-
30.	8 - 9	46,XY	-	-
31.	5 - 9	46,XX	-	-
32.	6 - 4	46,XX	-	-
33.	7 - 1	46,XX	-	-
34.	7 - 3	46,XY	-	-
35.	7 - 12	46,XX	-	-
36.	7 - 0	46,XY	-	-
37.	6 - 5	46,XY	-	-
38.	7 - 8	46,XY	-	-
39.	7 - 5	46,XY	-	-

TABLE 34 (continued)

Case no.	Birthweight (lbs. - oz.)	Chromosome analysis	Biochemical abnormality	Family history
40.	8 - 4	46,XX	-	-
41.	8 - 0	46,XX	-	-
42.	7 - 0	46,XX	-	-
43.	7 - 15	46,XY	↑muco.	+
44.	unknown	46,XX	-	-
45.	3 - 15	46,XY	-	-
46.	7 - 10	46,XY	-	unknown
47.	8 - 0	46,XX	-	-

TABLE 34 (continued)

Case no.	Stigmata
1.	high arched palate, seizures, varus of heel
2.	seizures, increased carrying angle
3.	high arched palate, scoliosis, epicanthus, pectus excavatum
4.	r. cryptorchidism, low ant. hairline, pectus excavatum, broad nose
5.	high arched palate, strabismus
6.	endophthalmus, blind
7.	misshapen ears
8.	broad nose, endophthalmus, protruding tongue, clinodactyly
9.	high arched palate, misshapen ears, broad nose, antimongoloid slant, cryptorchidism, short little fingers
10.	dolicheocephaly, synophoris, arachnodactyly, broad nose, bulbous great toes
11.	scoliosis, seizures
12.	high arched palate, heart defect
13.	seizures
14.	strabismus, microphthalmia
15.	broad nose, prognathia, juvenile ext. genitalia
16.	high arched palate, microcephaly, exophthalmus
17.	none
18.	high arched palate, cryptorchidism, prognathia
19.	scoliosis
20.	cryptorchidism, hypospadias, dextrocardia, seizures
21.	l. eye microphthalmic, misshapen ears, facial asymmetry
22.	strabismus, epicanthus, broad nose
23.	none
24.	r. cryptorchidism, high arched palate
25.	strabismus, endophthalmus
26.	none
27.	prognathia
28.	high arched palate, frontal bossing
29.	synophoris
30.	high arched palate, prognathia, misshapen ears, endophthalmus
31.	high arched palate, frontal bossing, scoliosis, enlarged clitoris and underdeveloped labia, broad nose
32.	strabismus, endophthalmus, absent eyelashes, webbed neck, heart murmur, increased carrying angle
33.	high arched palate, misshapen ears, seizures, epicanthus, facial asymmetry, antimongoloid slant, hirsute

TABLE 34 (continued)

Case no.	Stigmata
34.	high arched palate, misshapen ears, ptosis, low anterior hairline
35.	high arched palate, blind, microcephaly, seizures, microcephaly, nystagmus, increased carrying angle
36.	none
37.	cryptorchidism, broad nose
38.	high arched palate, strabismus, misshapen ears, cryptorchidism, supernumerary digit
39.	none
40.	none
41.	none
42.	microcephaly, strabismus
43.	broad nose, misshapen ears
44.	endophthalmus
45.	high arched palate, antimongoloid slant, facial asymmetry, cryptorchidism, microcephaly, ptosis, epicanthus
46.	high arched palate, endophthalmus, scoliosis
47.	brachycephaly, low posterior hairline

TABLE 35. Relationship between level of mental retardation and the number of physical abnormalities among individuals with an arch or radial loop on at least one number I digit

Level of retardation	0	1	2	3 or more	total
profound	2	3	4	13	22
severe	1	3	7	7	18
moderate	3	0	2	1	6
mild	1	0	0	0	1
total	7	6	13	21	47

(5/47). A positive correlation was seen between progressively severe retardation and the number of physical abnormalities (Table 35).

Because of the high frequency of cryptorchidism observed among the males of this group, these eight individuals with cryptorchidism were re-examined to determine if any other physical stigmata were common to them. Except for the presence of high arched palates in five of the eight and broad noses in three of the eight, no such similarities were observed. Subsequently, one of the eight was seen to have a biochemical abnormality (case number 14), and one was seen to have a chromosome abnormality (case number 4).

Family and biochemical data

Parental ages at the time of birth were available for 43 of the 47 individuals. The mean paternal age was 30.9 ± 6.6; the mean maternal age was 27.1 ± 5.9. However, among parents of four individuals with chromosome abnormalities, the mean parental ages were increased (mean paternal age = 35.3; mean maternal age = 35.0).

Birthweights were available for 43 of the 47 individuals, including four reported as premature (cases 2, 6, 14, 45). The mean value for 26 males was 7 lbs. 1.3 oz. ± 19.4 oz.; the corresponding value for 18 females was 6 lbs. 11.3 oz. ± 25.4 oz.

Examination of family histories with respect to occurrence of mental retardation revealed positive findings for five of the 47 families (cases 8, 9, 25, 28, 43). Pedigrees for these families are shown in Figure 7. Attempts to initiate family studies were largely unsuccessful, due either to death or unavailability of informative family members. The sister of case 25, however, was examined for possible dermatoglyphic abnormalities and other physical stigmata. Like her brother, she was seen to be profoundly retarded (I.Q. = 20; Stanford-Binet) and exhibited endophthalmus. Furthermore, dermatoglyphic analysis revealed a radial loop on her left fourth digit (ten-finger pattern - UUUUU UUURU); while this is a finding dissimilar to that of her brother, it must nevertheless be considered as an abnormality, since the presence of a radial loop on digit 4 is rare among normals. The maternal uncle of case 43 was also examined; he exhibited no dermatoglyphic, physical, or biochemical abnormalities.

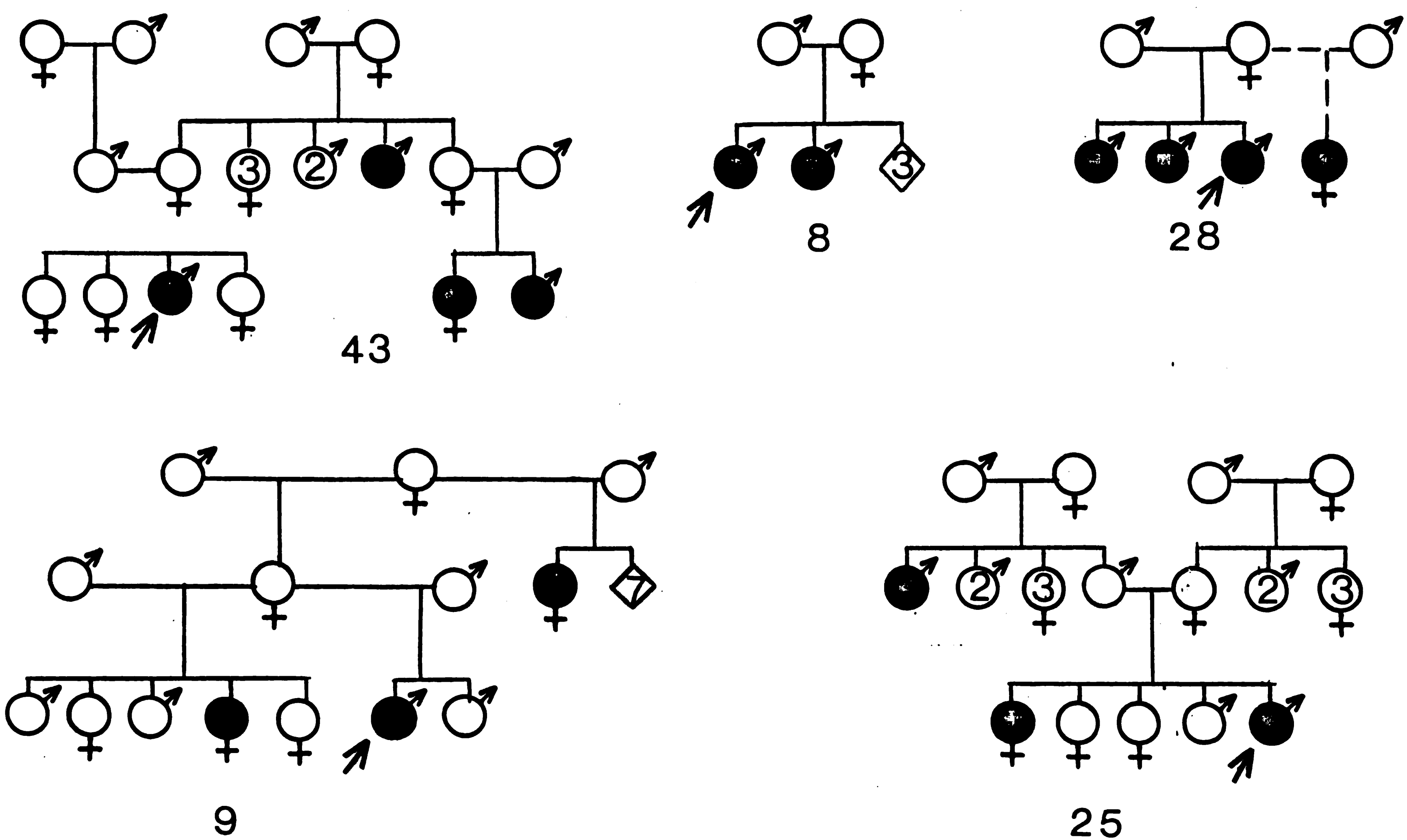

FIGURE 7. Family pedigrees from five retarded individuals with abnormal thumbprints whose family histories were positive for mental retardation

Results of serum and urine screens obtained from the records of the 47 individuals showed abnormalities in three of the individuals (6.4%). Two of the three individuals tested positive for mucopolysaccharides (cases 9 and 43), and the third showed an increase in phenylalanine excretion. On physical examination, however, the first two individuals showed no characteristics typical of any of the mucopolysaccharides; nor did the third individual display any features characteristic of phenylketonuria.

Family histories of all three individuals were positive with respect to occurrence of mental retardation (Figure 7). However, attempts to study the families further were unsuccessful.

Cytogenetic evaluation

Forty-three of the 47 individuals examined were seen to have normal chromosome constitutions (25 normal males; 18 normal females). Four individuals were observed with chromosome abnormalities, all involving acrocentric chromosomes. Cases I and II each had an additional chromosome which was bisatellited and apparently dicentric. Case III had an extra band on a chromosome 15. Case IV was seen to have an extra chromosome, approximately the same size as a G-group chromosome. Case reports and detailed cytogenetic descriptions of these four individuals follow.

Case I

The propositus is a 17 year old severely retarded Caucasian male. He was born to a 38 year old, gravida 4, para 3 female, and a 38 year old male. Both the propositus' sibs are phenotypically normal. The pregnancy was complicated by severe vaginal bleeding at three months which lasted one week. Otherwise the pregnancy was uneventful, and there is no report of any hazardous exposure, infection, or unusual medication. Delivery was at full term and normal; birth weight was 7 lbs. 14 oz. Except for an unusually shrill cry, no unusual features were noted at birth or in the neonatal period. The propositus sat at 10 months and walked at 15 months; he has never acquired speech. At age 5 he was dismissed from a special education class because of hyperactivity and destructive behavior; he was admitted to a mental retardation institution at age 9. At age 10 he was reported to have had a seizure, and was hospitalized for observation. No seizures were observed, and an EEG taken at that time revealed no obvious abnormalities. Over the next few years, however, the propositus was reported as having several seizures, and an EEG taken at age 16 revealed a generalized dysrhythmia. The propositus is presently receiving anti-convulsive medication daily.

Physical examination revealed a severely retarded male (I.Q.=24; Stanford-Binet) with few obvious physical abnormalities (Figure 8). Height (65 inches), weight (114 lbs.), and head circumference (21 1/4) were all in the low normal range. Observed physical stigmata included a high arched palate, broad nasal bridge, and minimal varus of the heel. Previous reports had indicated strabismus, but this was not apparent. No other unusual features were noted. Urinalysis and C.B.C. values were normal.

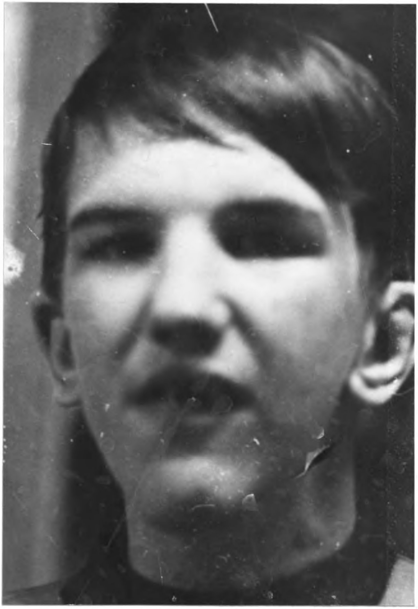

FIGURE 8. Case I

Cytogenetics

Giemsa-trypsin banded preparations revealed an additional acrocentric chromosome midway in size between a D and G group chromosome (Figures 9 and 10). The extra chromosome was present in all cells examined. Two bands were consistently seen on the additional chromosome, one in the centromeric region and one at the distal end of the long arm. This banding was consistent with that of a deleted chromosome 15 with a break through, or just distal to, band q21 (Paris Conference, 1971). Therefore, a tentative diagnosis of partial trisomy for the proximal segment of chromosome 15 seemed reasonable. However, a subsequent examination of quinacrine banded preparations revealed the additional chromosome to be bisatellited, suggesting a more complex rearrangement. This was confirmed by sequential quinacrine to centromeric banding, in which the extra chromosome was seen to have two distinct centromeric bands, corresponding to the two bands seen with giemsa-trypsin banding (Figure 11).

Because of the known instability of dicentric chromosomes (Sears and Camara, 1951), attempts were made to demonstrate the presence of an additional cell line. However, evaluation of 100 cells from three different blood cultures showed no evidence for mosaicism. The dicentric chromosome was occasionally missing from a division, but this was apparently due to random loss as its frequency (7/100) was approximately the same as the other acrocentrics.

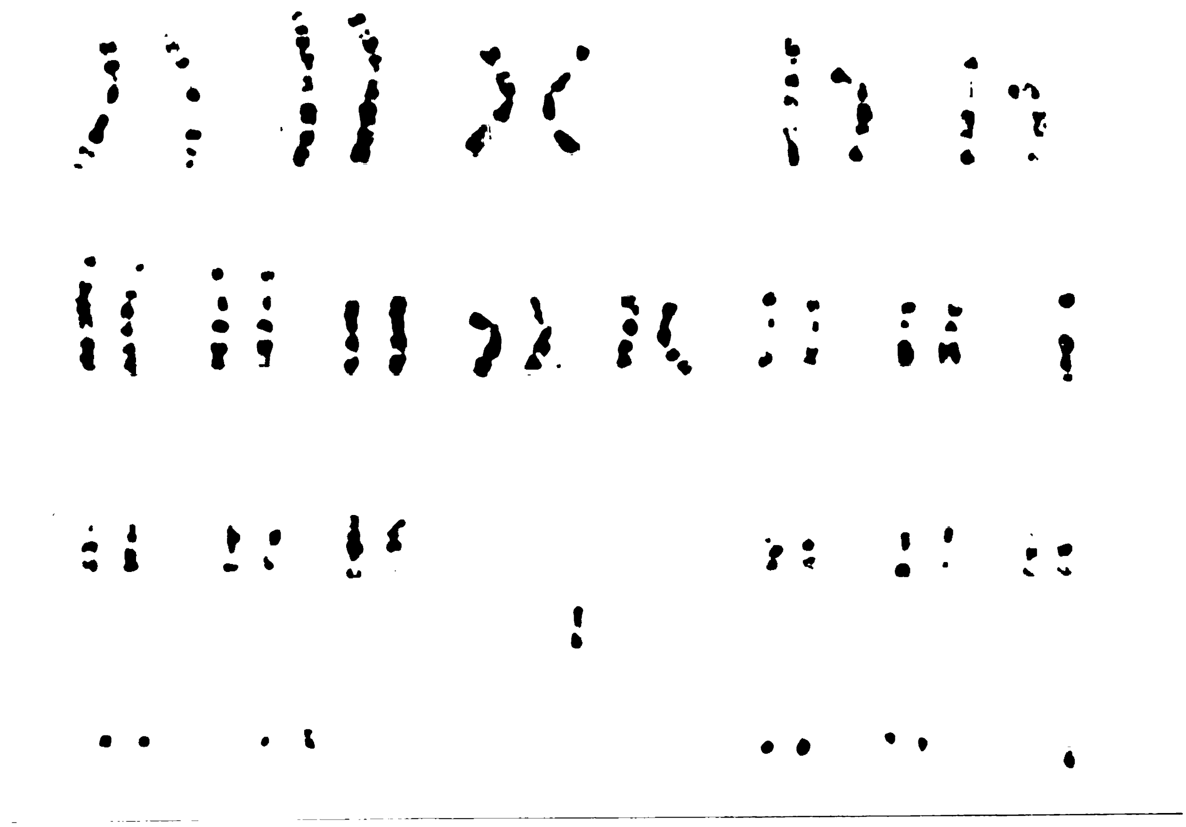

FIGURE 9. Giemsa-trypsin banded karyotype of Case I

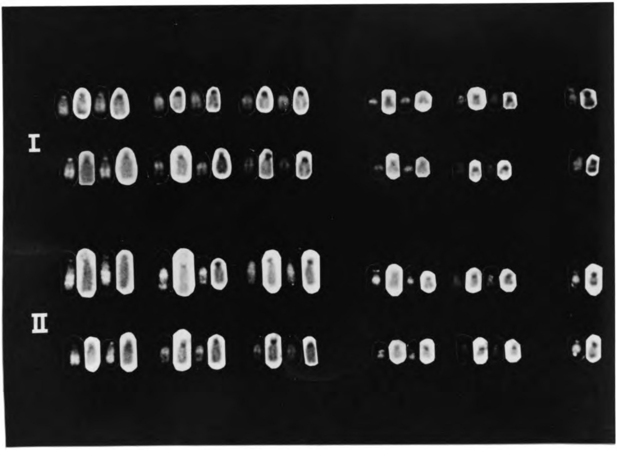

FIGURE 10. Partial karyotype of cases I and II, using sequential Q to C banding

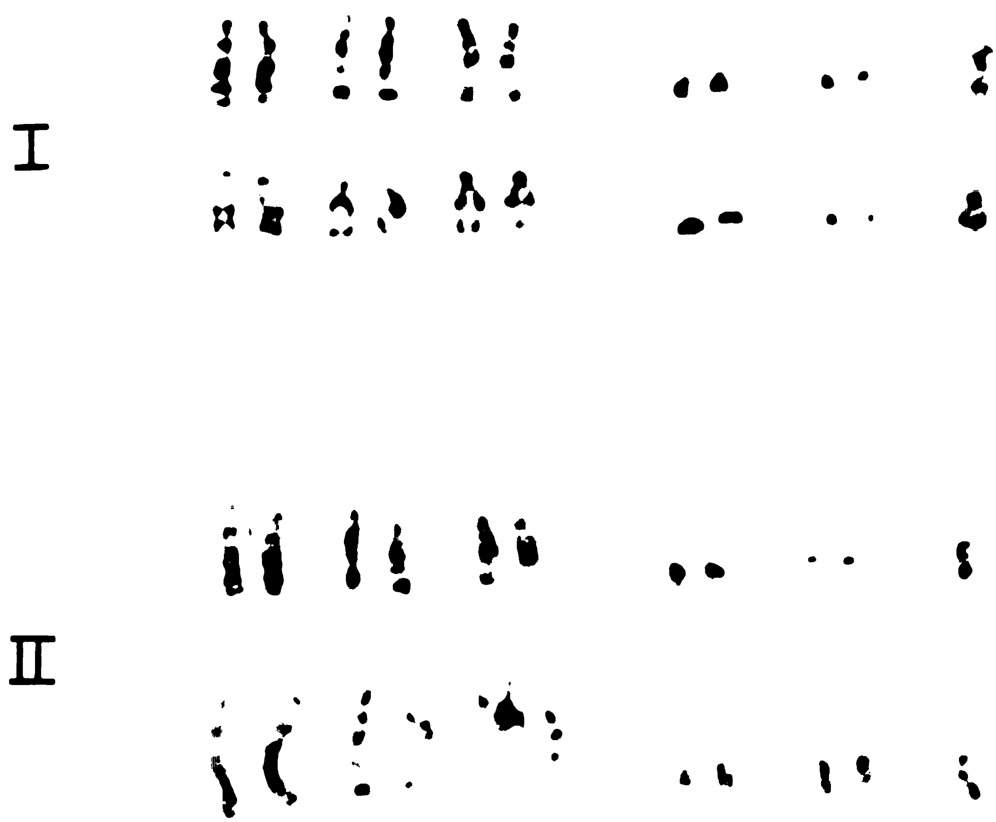

FIGURE 11. Partial giemsa-trypsin banded karyotypes of cases I and II

Other studies were carried out to determine whether one or both of the centromeres was regularly constricted. This was facilitated by the fact that one of the two pairs of satellites consistently stained more intensely than did the other (Figure 11), allowing easy discrimination between the two positively staining C-band regions. Analysis of the two centromeres showed that only the centromere closest to the less intensely staining satellites was ever constricted.

Examination of blood cultures from the propositus' parents revealed no obvious abnormalities. Attempts to use chromosome heteromorphisms in order to specify the parental origin of the extra chromosome were unsuccessful. Dermatoglyphic analysis of the parents revealed no unusual findings in the mother; the father, however, had arches bilaterally on the number 1 digits.

<u>Case II</u>

The proposita is a 48 year old, severely retarded female. She was the product of a pregnancy between a 30 year old, gravida 3, para 2 female, and a 30 year old male. The couple subsequently had another child three years later; both of the proposita's sibs are phenotypically normal and have normal children themselves.

The pregnancy was complicated by two instances of hemorrhaging during the first trimester, and an acute gall bladder infection combined with bronchitis during the seventh month. The delivery was normal and was reported as one month premature. Birth weight was 6 lbs. 8 oz. No unusual features were noted at birth or in the early neonatal period.

At ten weeks of age, the proposita became ill with what was diagnosed as influenza, and suffered a period of convulsions lasting two days. She first sat at 13 months, walked at 24 months, and talked at 3 years. She was admitted to an institution for the mentally retarded at age 6. On admission and over the next few years, she was reported as being hyperactive and destructive. Menses began at 14 and were normal. Major motor seizures began at age 18 and occurred frequently (averaging approximately one per month) over the next 12 years. An EEG taken at 39 showed spiking; subsequent EEG's taken at 45 and 46 were interpreted as having no obvious abnormalities. The seizures have now been controlled with medication, and no episode has been reported in the last 10 years. The proposita presently functions in the severe range of retardation, with an I.Q. of 24 (Stanford-Binet).

Physical examination revealed few obvious abnormalities (Figure 12). She is short (59 inches), but values for weight (112 lbs.) and head circumference (22 1/2 inches) were within normal limits. Unusual physical features seen included multiple nevi of the face and chest, and an increased carrying angle at the elbow. The distal portion of the third digit on the left hand was bulbous and the nail hypoplastic, but this may have resulted from an injury. No other stigmata were noted. Urinalysis and C.B.C. values were normal.

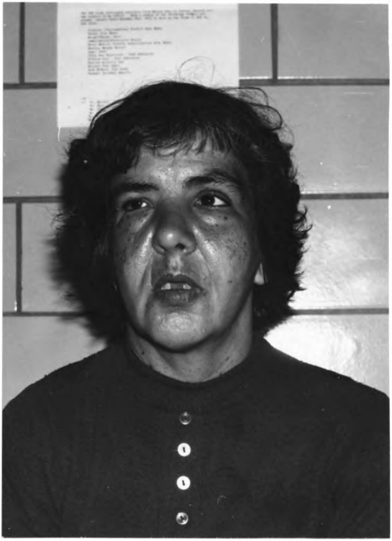

FIGURE 12. Case 11

Cytogenetics

Giemsa-trypsin banded analysis revealed an additional acrocentric chromosome intermediate in size between a D and G group chromosome. A partial G-banded karyotype of the proposita's acrocentric chromosomes, including the extra chromosome, is given in Figure 10. The additional chromosome was seen in all cells examined. In most preparations two bands were seen, one at the apparent centromere at one at the distal end of the long arm. However, in some early metaphase or late prophase divisions an additional, faint, band was seen close to the apparent centromere. Analysis with quinacrine banding showed the extra chromosome to be bisatellited. Subsequent quinacrine to centromeric banding showed two bands, corresponding to the two obvious bands seen with giemsa-trypsin banding.

Two repeat samples were analyzed, with no indication of mosaicism (the extra chromosome was seen in 95 of 100 cells examined from the three cultures). Examination of the two centromeric bands (with centromeric banding) showed that only the one closest to the faint G-band was regularly constricted. Karyotypes of the proposita's mother, sister, and brother were all normal; the father is no longer living. Dermatoglyphics of the proposita's mother and sister were not unusual.

Case III

The propositus is a 20 year old profoundly retarded male with multiple congenital abnormalities. He was the seventh born in a sibship of ten. Maternal and paternal ages at the time of birth were 35 and 34, respectively. There is no history of spontaneous abortions in the family, and all the propositus' sibs are reported as phenotypically normal. The pregnancy was uneventful and was complicated only by exposure to X-rays during the fifth month. The delivery was at term and normal, and no unusual features were noted at that time. Birthweight was 7 lbs. 2 oz.

Early development was not markedly delayed. The propositus sat at eight months and walked at one year. However, he is still not toilet trained and has no speech. He was admitted to an institution for the mentally retarded at age seven.

Physical examination revealed a small, retarded male (I.Q. = 18; Catell) with multiple abnormalities (figure 13). Height was 60 1/2 inches, weight 110 lbs., and head circumference 22 inches. Observed physical abnormalities include a slightly asymmetric face, high arched palate with malocclusion, minimal pectus excavatum, pronounced thoracic kyphosis, bilateral syndactyly of the second and third toes, hyperactive deep tendon reflexes, and bilateral positive Babinski signs. An EEG taken at age 5 showed a paroxysmal disturbance in the right posterior hemisphere; however, he has never had a seizure. Urinalysis and C.B.C. values were normal.

Cytogenetics

Analysis of giemsa-trypsin banded metaphases revealed that the long arm of one of the chromosome 15's was consistently longer than

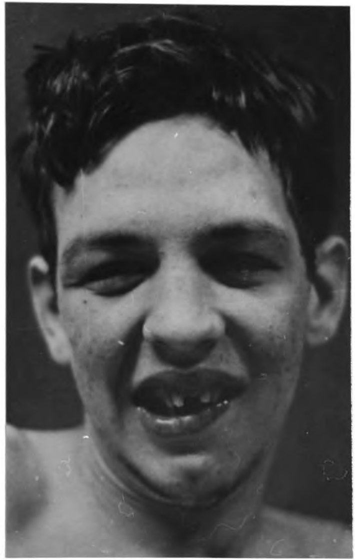

FIGURE 13. Case III

its homologue (Figure 14). A comparison of the two homologues showed that the distances from the beginning of band q21 to the centromere, and from the end of band q21 to the distal end of the long arm, were approximately the same for both. However, band q21 itself (and perhaps some of the bordering interband material) is apparently present in duplicate on the abnormally long chromosome 15.

Examination of the parental karyotypes revealed no abnormalities in either one. Dermatoglyphics of the propositus' parents were not unusual, except that the mother had an ulnar loop with a low ridge count on her left first digit.

Case IV

The propositus is a 25 year old severely retarded male. He was born to a 33 year old female and a 35 year old male, and was their first child. The couple subsequently had three more children, all of whom are reported as normal and in good health. Additionally, three spontaneous abortions have been reported, all occurring at about 2 1/2 months; information concerning the positioning of these abortions with respect to the other pregnancies was not available. The pregnancy was reported as normal. The mother received vitamins and medication for weight control; otherwise, no unusual complications were reported. The delivery was at term, and forceps were used. The birthweight was 7 lbs. 14 oz. The propositus was kept in the hospital for four weeks following birth, but information regarding the reason for this was not available.

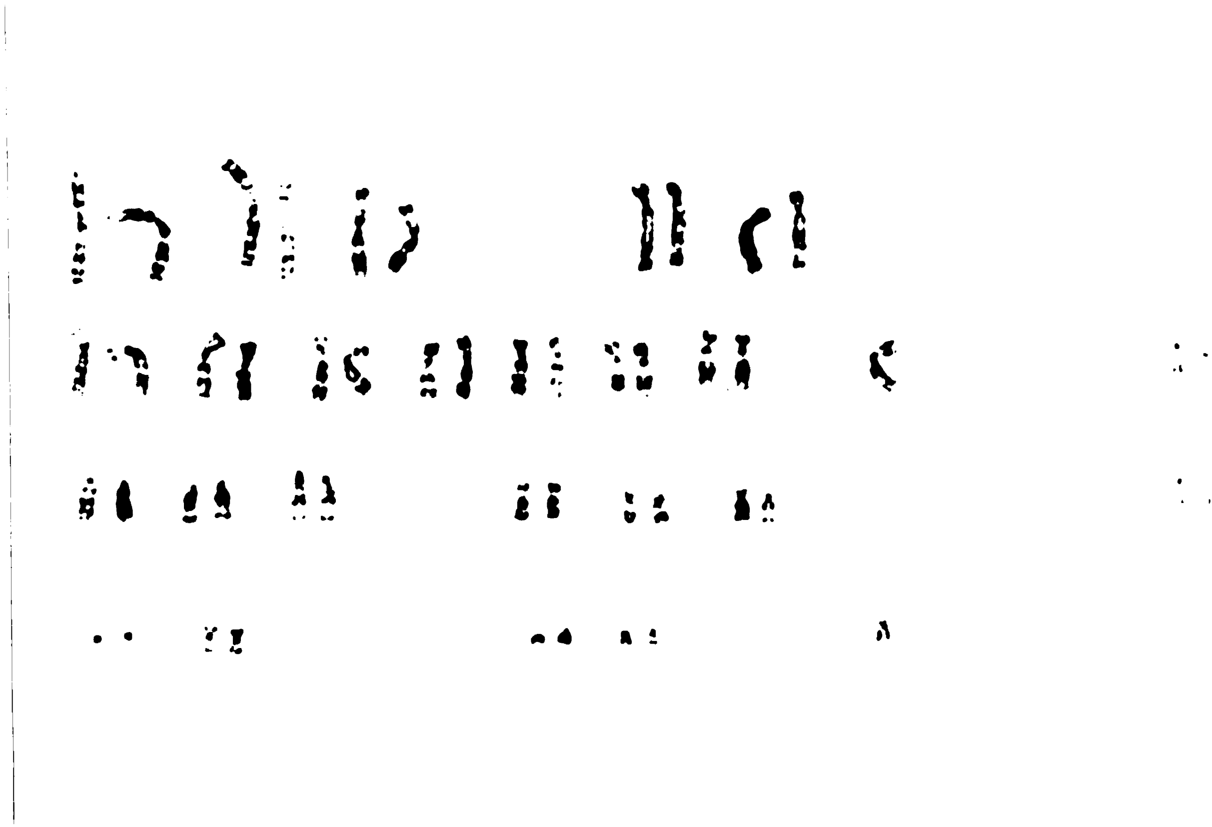

FIGURE 14. Giemsa-trypsin banded karyotype of case III; the four chromosomes at the right are two pairs of #15's from two additional cells

Developmental milestones were not markedly delayed. The propositus sat at seven months, walked at 11 months, and was toilet trained at two years. However, he did not begin to speak until age two years, and still uses only a few words. The propositus was admitted to an institution for the mentally retarded at age 13. He presently functions in the severe range of mental retardation, with an I.Q. of 31 (Stanford-Binet).

Physical examination revealed a severely retarded male with multiple anomalies. Height was 68 1/2 inches, weight 143 lbs., and head circumference 22 inches. Abnormal facial features included a low anterior hairline, a bulbous nose, and endophthalmus. Other stigmata included pronounced pectus excavatum, and an undescended right testis. Urinalysis and C.B.C. values were normal.

Cytogenetics

Analysis of giemsa-trypsin banded metaphases revealed an additional chromosome, approximately the same size as a G group chromosome, in all divisions examined (Figure 15). However, the banding pattern of the extra chromosome was not consistent with either a chromosome 21 or 22. Subsequent analysis using quinacrine banding definitely showed the additional chromosome to be satellited, indicating an acrocentric origin. Attempts to elucidate the origin of the extra chromosome were not successful. The parents were not available for cytogenetic evaluation.

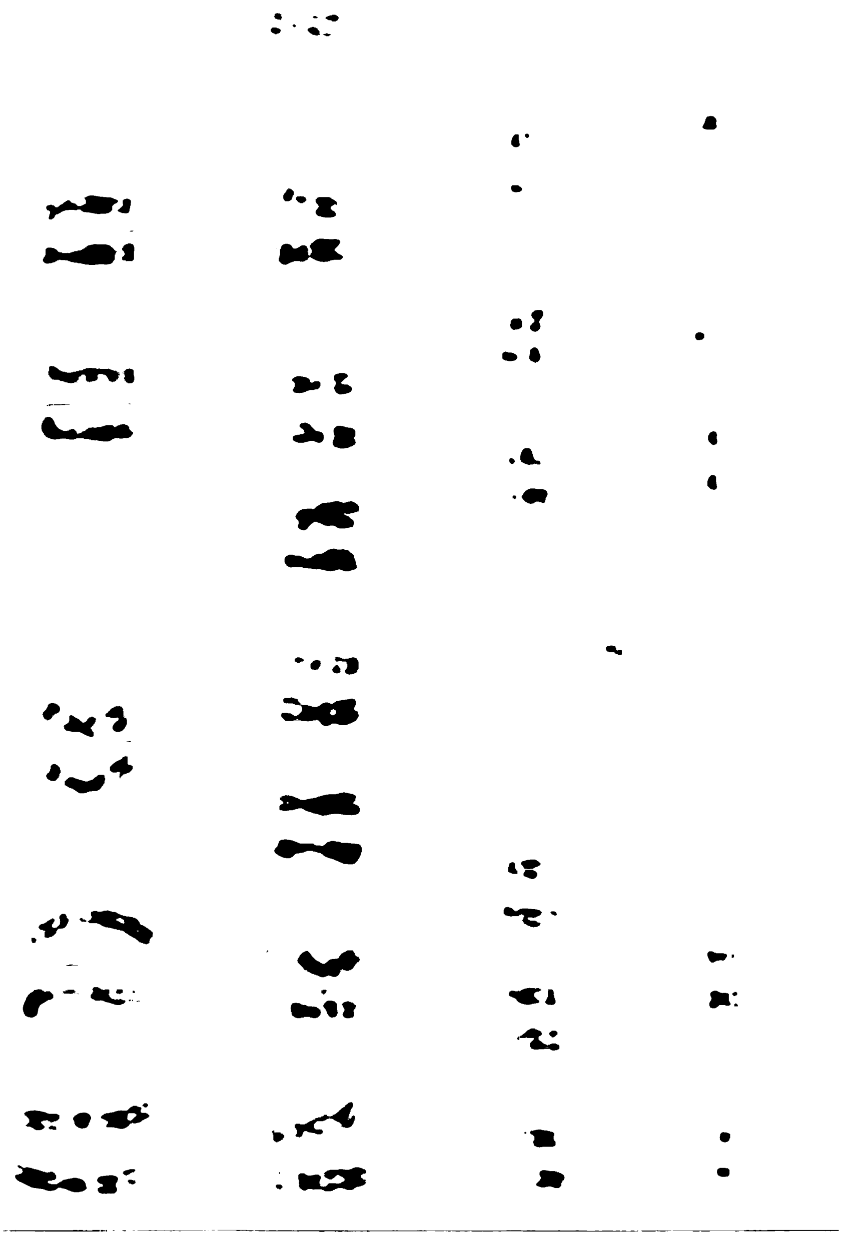

FIGURE 15. Giemsa-trypsin banded karyotype of case IV

DISCUSSION

Results of previous dermatoglyphic studies of control Caucasian populations are summarized in Table 36. Mean values for total finger ridge count and pattern intensity are in good agreement with those values found in the present study (Figures 3, 4, 5, 6). Additionally, the frequency distributions of the four digital patterns closely approximate those of the present study (Table 11).

Previous studies of fingerprint patterns among non-specific mentally retarded individuals have dealt primarily with frequency distributions of the four pattern types, and have yielded contradictory findings. The early studies of Bonnivie (1927) and Blumel and Poll (1928) showed decreases in whorls, on at least some of the digits, among the retarded individuals. The studies of Lu (1968) and Saksena and Mathur (1974), however, found an overall increase in both whorls and arches, and a decrease in ulnar loops. In the study of Alter (1966), no significant differences were noted between the control and retarded individuals. He did, however, find a slight increase in whorls and an accompanying decrease in ulnar loops among both the male and female retarded groups.

In the present study, an analysis of 1463 control individuals and 868 mentally retarded individuals revealed the following significant differences between the control and retarded groups:

TABLE 36. Normal values for total finger ridge count, pattern intensity, and frequency of digital pattern types among Caucasian individuals

Total finger ridge count (mean values)[1]

males (n=1519)	females (n=1523)
141.30	124.94

Pattern intensity (mean values)

males	females[1]
11.8	11.4

males and females[2] (n=5000)
12.1

Frequency distribution of digital pattern types[3]

	males (n=358)	females (n=358)
ulnar loop	.619	.645
whorl	.288	.264
radial loop	.052	.037
arch	.040	.054

[1]Holt (1968) [2]Cummins (1943) [3]Plato et al. (1975)

TABLE 36 (continued)

Frequency distribution of pattern types on the ten digits[3]

Males (n=358)

	Right Hand 1	2	3	4	5	Left Hand 1	2	3	4	5
Ulnar loop	.555	.324	.740	.522	.813	.668	.369	.710	.631	.868
Whorl	.431	.349	.187	.455	.179	.299	.332	.173	.346	.137
Radial loop	.000	.215	.022	.014	.000	.003	.232	.028	.006	.000
Arch	.015	.112	.050	.008	.008	.031	.067	.089	.017	.006

Females (n-358)

	Right Hand 1	2	3	4	5	Left Hand 1	2	3	4	5
Ulnar loop	.598	.413	.810	.570	.855	.635	.363	.735	.609	.869
Whorl	.372	.330	.126	.400	.131	.315	.332	.162	.358	.109
Radial loop	.000	.134	.014	.020	.000	.003	.173	.014	.011	.000
Arch	.031	.123	.050	.011	.014	.048	.131	.089	.022	.022

[3]Plato (1975)

1. among the retarded males, there was an overall increase in the frequency of arches and a decrease in whorls
2. among the females, there was an increase in both arches and whorls, and a decrease in ulnar loops.

If the findings of the males and females are combined for both the control and retarded groups, three differences are seen between the control and retarded groups - an overall increase in both arches and whorls, and a decrease in ulnar loops. This finding is in good agreement with the reports of Lu (1968) and Saksena and Mathur (1974), for which males and females were analyzed together. However, it does not coincide with the findings of Bonnevie (1927), Blumel and Poll (1928), or Alter (1966). In part, the differences seen between the present study and these three studies may be related to differences in the populations sampled. The studies of Bonnevie (1927) and Blumel and Poll (1928) were not controlled with respect to type of retardation, and may have included individuals who would have been screened from the present study (e.g., Down's syndrome, which is known to be associated with decreased frequency of whorls). However, differences seen between the present study and that of Alter (1966) are not easily explained, since in that study all individuals were screened to rule possible genetic or environmental disorders. It is possible that these differences may be partly due to the changing structure of the institutional population in Michigan during the time of the present study (i.e., increasing proportion of more severely retarded individuals); this seems unlikely, however, in light of the fact that little difference was seen between the various levels of retardation with respect to distribution of the four pattern types. Alternately, the differences between Alter's study and the present study may be partially a reflection of the differences in sample sizes. Alter's study included analysis of only 200 control individuals (100 of each sex) and 376 retarded individuals (213 males, 163 females). With respect to the rarely occurring patterns (i.e., radial loops and arches), such a small sample size may be inadequate to accurately determine whether differences exist between the control and retarded groups.

Fingerprint pattern combinations in excess among mentally retarded individuals

Examination of fingerprint pattern distribution and pattern combinations led to the detection of 18 types of ten-finger patterns in excess among the retarded individuals of either or both sexes (Table 32). Such patterns fall into two general categories: those in which a rarely seen pattern type (i.e., an arch or radial loop) is found either in increased frequency or on a digit other than expected, and those in which an unusual pattern combination is found.

In the former category, almost all ten-finger patterns seen in excess among the retarded individuals involve the presence of one or more arches. The occurrence of arches has been related to a weak volar pad development (Hirsch and Schweichel, 1973), and excess numbers of arches have been seen in association with both genetically and environmentally caused disorders (Alter, 1966). Thus it seems likely that the excess of arches (or unusual location of arches) seen among the mentally retarded individuals result from heterogeneous developmental disturbances affecting volar pad development.

However, a comparison of I.Q. levels among retarded individuals with such ten-finger patterns suggests the possibility of a specific factor related to arch production. For while no obvious differences were seen between the different levels of retardation with respect to total finger ridge count, pattern intensity, or overall frequency of the four pattern types, there is an increase in the frequency of profoundly or severely retarded males for almost all ten-finger pattern categories involving arches (Table 33).

One possible explanation for the observed difference in these ratios would be the presence of X-linked mental retardation disorders associated with profound or severe mental retardation and changes in fingerprint patterns. Under such a scheme, the presence of arches in ten-finger patterns of retarded individuals would result from one of three mechanisms:

1. normal familial transmission of such patterns
2. autosomal errors (either single gene or chromosomal) or environmental factors leading to the production of such ten-finger patterns; such factors would account for the increase of these patterns in the retarded females and moderately or mildly affected males, and for part of the increase among the more severely affected males

3. X-linked mental retardation associated with severe or profound retardation and fingerprint abnormalities; this would account for the further increase in such patterns seen among the more severely affected males.

Studies of fingerprint patterns in families apparently positive for X-linked retardation would be instructive in determining the validity of this thesis. For such families, it would be expected that the affected males would have a higher frequency of arches than would their normal sibs. Unfortunately, in the present study there is little data that would serve to test this hypothesis; of the five families with positive histories for mental retardation, only one appears consistent with an X-linked mechanism (Figure 7).

Factors related to the production of unusual pattern combinations among the mentally retarded individuals (i.e., the second category of ten-finger patterns seen in excess among the retarded individuals) are not readily apparent. Among the males, the increased occurrence of ulnar loops bilaterally on the number I digits may, in part, be related to disturbances in volar pad development, since the resultant pattern combinations involve less complex patterns. Among the retarded females, however, those ten-finger patterns seen in excess generally involve an increase in more complex patterns, and cannot be explained by the same mechanism. Factors related to the production of such factors are not obvious.

Examination of individuals with a radial loop or arch on at least one number I digit

Forty-seven mentally retarded individuals with an arch or radial loop on at least one number I digit were examined in an attempt to discover similarities (with respect to family histories, possible biochemical or chromosomal abnormalities, and physical stigmata) indicative of mental retardation syndromes. Mean paternal and maternal ages at birth were 30.9 ± 6.6 and 27.1 ± 5.9, respectively; these are in good agreement with corresponding values seen in the control Caucasian population (mean paternal age = 30.7; mean maternal age = 28.7; Hamerton, 1971). The mean birthweight for the retarded males in this group was 7 lbs. 1.3 oz. ± 19.4 oz. and for females was 6 lbs. 11.3 oz. ± 25.4 oz. These values are somewhat lower than the corresponding control values (males- 7 lb. 8 oz.; females - 7 lb. 4 oz.; Altman and Dittmar, 1962).

Upon physical examination, 40 of the 47 retarded individuals (85.1%) were felt to have one or more abnormal physical features. This frequency is much greater than that noted by Hanefeld (1972), who found abnormal clinical findings in only 93 of 163 non-specific mentally retarded individuals (57.1%). The most frequent clinical findings in Hanefeld's sample were epilepsy (24%), microcephaly (10.4%), and hydrocephaly (8.5%). In our study the most frequently seen abnormalities were high arched palate (38.3%), malformed ears (17.0%), cryptorchidism (28.6% of males), and endophthalmus (14.9%); seizures occurred in 12.8% of the 47 individuals, microcephaly in

8.5%, and hydrocephaly was not seen. Differences between our study and Hanefeld's may reflect real biological differences between the two groups of individuals studied, but are probably also partly due to differing definitions of what constitutes an abnormal clinical finding. However, two abnormal findings (i.e., cryptorchidism and chromosome abnormalities) were seen in such high frequencies in the present study that they are considered separately below.

Incidence of cryptorchidism

Bilateral or unilateral cryptorchidism is seen in less than 1% of all males over 12 months of age (Beeson and McDermott, 1967). However, among 28 retarded males with an arch or radial loop on a number 1 digit, 8 (28.6%) were seen with either bilateral or unilateral cryptorchidism. This frequency is also higher than that seen among retarded males in general. Of 130 retarded males randomly selected from the Oakdale Center, only 7 (5.4%) had cryptorchidism. The difference between the randomly selected group and the group ascertained through abnormal thumbprint patterns was found to be significant at p=.01 (Table 37). This may actually underrepresent the difference between these two groups, since the fingerprint patterns of the randomly selected group were not known beforehand, and some of the individuals with cryptorchidism conceivably may also have had abnormal fingerprint patterns (in fact, two of these individuals were subsequently examined dermatoglyphically; one was found to

TABLE 37. 2x2 contingency table to test the hypothesis that differences in frequency of cryptorchidism exist between mentally retarded males with cryptorchidism and mentally retarded males in general

Observed:

	Cryptorchidism	No cryptorchidism	Total
randomly selected males	7	123	130
males with abnormal thumbprint	8	21	29
	15	144	159

Expected:

randomly selected males	12.26	117.74
males with abnormal thumbprint	2.74	26.26

Chi-square = 2.26 + .23 + 10.10 + 1.05 = 13.64

Chi-square $_{.01,1}$ = 6.63

have a radial loop on a number 4 digit, a configuration rare among normals).

Subsequent biochemical and cytogenetic evaluation revealed that two of the eight individuals with cryptorchidism and abnormal fingerprint patterns on digit I also had biochemical or chromosome abnormalities; these errors may presumably have been causally related to the cryptorchidism. Among the remaining six individuals, for whom no such abnormality was apparent, certain additional dermatoglyphic similarities were found. Four of the six individuals had only one arch (that one on the number I digit), compared with only four of the 20 males without cryptorchidism. Additionally, these same four individuals all had at least one whorl, and always on at lease one number 2 digit; among the 20 males without cryptorchidism, however, only seven had one or more whorls and in only two individuals were whorls seen on digit 2.

These findings (i.e., occurrence of arches only on digit I and the occurrence of more complex patterns on the other digits) are suggestive of a developmental process affecting only the fingerprints on digit I. At least one other such syndrome is known - Holt-Oram syndrome. In this disorder, fingerprint patterns on digits other than digit I appear to be little different from those in control individuals. However, the fingerprints on digit I are markedly abnormal in that approximately 50% of the individuals with this syndrome have a radial loop on at least one number I digit (Hassold, unpublished date). However, abnormalities of forelimb development which are often seen in association with Holt-Oram syndrome were not detected in any of the individuals with cryptorchidism. Furthermore, a subsequent examination of these six individuals failed to show any physical similarities suggestive of a common etiology for their retardation. So while the data strongly suggest a correlation between abnormal fingerprint patterns on digit I and cryptorchidism, no one causative agent for such abnormalities could be detected.

Incidence of chromosome abnormalities

Four of the 47 individuals (8.5%) had radial loops or arches on at least one number I digit and were seen to have abnormal chromosome constitutions. All abnormalities involved autosomal acrocentric chromosomes.

The frequency of chromosome abnormalities seen in the present study is compared to other cytogenetic surveys of mentally retarded individuals in Table 38. In five of the six surveys the ascertainment was total; i.e., individuals were not selected on the basis of multiple abnormalities or suspected chromosome abnormality, but were rather intended to represent the institutional populations in general. The final study (State of Michigan) included individuals ascertained because of suspected chromosome abnormalities; however, it has been included because the institutions from which the individuals were selected and the time period of the survey are both roughly the same as the present study. For all surveys cited, finding with respect

TABLE 38. Previous cytogenetic surveys of mentally retarded individuals*

Study	Number of Individuals	Sex chromosome Abnormalities	Autosomal Abnormalities	Total Abnormal	% Abnormal
Sutherland & Barholomew (1971)	36	3	0	3	8.3
Newton et al. (1972)	1151	10	10	20	1.7
Fujita and Fujita (1974)	58	2	1	3	5.1
Speed et al. (1976)	2770	31	33	64	2.3
Jacobs (ongoing)	296	2	3	5	1.7
State of Michigan (ongoing)	658	10	6	16	2.4
TOTAL	4969	58	53	115	2.2
PRESENT STUDY	47	0	4	4	8.5

* excluding autosomal trisomies 13, 18, 21

to trisomies 13, 18, and 21 have been excluded, since these were screened from the present project at its onset.

Comparison of the results of previous cytogenetic surveys and the present study shows a four-fold increase in chromosome abnormalities in the present study (this difference is significant at p=.01; chi-square = 7.90, 1 d.f.). Furthermore, when only autosomal errors are considered, this becomes an eight-fold increase. Four explanations can be given to account for the increased frequency of chromosome abnormalities in the present study:

1. The population of the previous studies differ in some way from that of the present study (i.e., with respect to the 868 individuals initially studied in the present study).
2. The increase is coincidental.
3. The 47 individuals examined cytogenetically are not representative of all mentally retarded individuals with arches or radial loops on digit 1.
4. The results reflect a real increase in the frequency of chromosome abnormalities among retarded individuals with an arch or radial loop on at least one number 1 digit.

It is difficult to determine whether the populations of the previous surveys substantially differ from that of the present study. All but two (Fujita and Fujita, 1974; Jacobs, ongoing) sampled primarily from Caucasian populations; however, little information is available regarding the level of retardation among the individuals sampled. The fact that the frequency of abnormalities seen in the

State of Michigan survey so closely agrees with those from the other surveys supports the idea that no substantive differences exist between the populations of the previous studies and that of the present one. The Michigan survey sampled from the same institutions and during the same time period as the present study; additionally, any bias in that survey would be expected to be in the direction of in increased frequency of chromosome abnormalities, since individuals were often ascertained on the basis of suspected chromosomal abnormalities.

A 2x2 contingency table comparing the frequencies of chromosome abnormalities in the present study with that of the Michigan survey reveals the difference to be significant at p=.05 (Table 39). While

TABLE 39. 2x2 contingency table to test the hypothesis that differences exist with respect to the frequency of chromosome abnormalities seen in the Michigan survey and among individuals with abnormal thumbprints seen in the present study

Observed:	Normal karyotype	Abnormal karyotype	Total
Michigan survey	658	16	674
Present study	43	4	47
	701	20	721
Expected:			
Michigan survey	655.30	18.70	674.00
Present survey	45.70	1.30	47.00
	701.00	20	721.00

Chi square = 6.17 (Chi-square$_{1,.05}$=3.84) the numbers involved in the present study are small, this finding supports the idea that chromosome abnormalities are increased among retarded individuals with arches or radial loops on digit I.

Of the initial 77 retarded individuals found to have an arch or radial loop on digit I, 47 were profoundly or severely retarded and 30 were moderately or mildly retarded. However, of the 47 individuals who were available for cytogenetic evaluation, only seven were moderately or mildly retarded. This finding argues that the increase in chromosome abnormalities is partly due to an increased frequency of more severely affected individuals among the final 47 individuals; in fact, a repeat of the analysis in Table 39, (assuming that no chromosome abnormalities would have been seen in the 30 individuals not cytogenetically examined) shows that the difference between the two studies does not reach significance (Chi-square = 2.12; $\text{chi-square}_{1,.05}=3.84$). However, even assuming that no chromosome abnormalities would have been seen in the 30 individuals not available for chromosome analysis, the frequency of abnormalities seen in the present study is still more than twice as great as that seen in the previous surveys listed in Table 38 (and the frequency of autosomal errors is over four times as great).

While it cannot be conclusively stated that retarded individuals with an arch or radial loop on digit I have a greater frequency of chromosome abnormalities than does the retarded population in general, at least two finding support this idea:

1. an increased frequency of such fingerprint patterns on digit I among individuals with a variety of autosomal abnormalities (Preus and Fraser, 1971; Lewandowski and Yunis, 1975)
2. an increased incidence of autosomal errors among retarded individuals with abnormal fingerprint patterns on digit I seen in the present study.

Together, these findings strongly suggest that there is an increased frequency of autosomal chromosome abnormalities (but probably not sex chromosome abnormalities) among mentally retarded individuals with an arch or radial loop on at least one number I digit.

Detection of a probable chromosome syndrome

Of the four individuals seen with abnormal chromosome constitutions, two (cases I and II) had similar - if not identical - abnormalities. Both had a supernumerary chromosome midway in size between a D and G group chromosome, and in both cases the extra chromosome was seen to be both bisatellited and dicentric. Furthermore, a comparison of the clinical manifestations of the two individuals revealed that both had seizures, but few other obvious abnormalities.

Because the banding patterns of the additional chromosomes of the two individuals were originally thought to be consistent with that of a deleted chromosome 15, a comparison was made of the cytogenetic and clinical findings of these individuals and previously reported cases of partial trisomy 15. The findings are summarized in Table 40. Reports of Rethore et al. (1973) and Fujimoto (1974) have not been included, since in the former the partial trisomy 15 was combined with monosomy 21 and, in the latter, the trisomy was for the distal segment of 15.

The 17 reports of partial trisomy 15 can be grouped into two categories: those involving a translocation, and those involving the presence of a supernumerary chromosome. In the former case, identification of the translocation products in a normal carrier made the diagnosis of partial trisomy 15 unequivocal. However, in the latter group, the diagnoses are less reliable, since they were based on the detection of a supernumerary chromosome with a banding pattern resembling that of a deleted chromosome 15.

Partial trisomies of several autosomal chromosomes are known to be associated with characteristic clinical pictures (Escobar et al., 1974; Escobar and Yunis, 1974; Lewandowski and Yunis, 1975). Thus it would be anticipated that the clinical manifestations would be similar for individuals with partial trisomy for the proximal portion of chromosome 15, regardless of the origin of the trisomic segment. However, a comparison of the two types of partial trisomy 15 listed in Table 40 shows that this is not the case. All five trisomic individuals whose diagnoses were verified through a translocation display mental retardation in association with a large number of physical abnormalities. However, among those individuals with a supernumerary chromosome only two individuals (those of Magenis et al. and Mankinen et al.) display such a variety of stigmata; one of these (Magenis et al.) has several abnormalities seen also among

TABLE 40. Previous reports of partial trisomy 15

Study	Chromosome constitution	Trisomic portion	M.R.	seizures	dermatoglyphic abnormality	bisatellited	Other stigmata
Parker & Alfi (1972)	47, XX +15q-	pter-q22	+	+			strabismus, hyperactive, immature speech, thin helices of ears
Magenis et al. (1972)	47, XX +15q-	p&q proximal	+				strabismus, growth retardation, epicanthus, broad nose, large mouth with full lips, antimongloid slant, separated teeth, large ears, thoracic kyphosis, increased carrying angle, spindle shaped fingers, short little fingers
Bucher et al. (1973)	47, X? +15q-	p&q prox.	+		-		esotropia, slender, hyperactive
Crandall et al. (1973) a.	47, XY +15q-	pter-q22	+	+	-	+	epicanthal folds, hypotonia, slight mongloid slant
b.	47, XY +15q-	pter-q22	+	+	-	+	endopthalmus, strabismus, high arched palate, hypotonic
Wurster-Hill & Hoefnagel (1973)	47, XY +15q-	pter-q23	+	+	+	+(?)	high arched palate, abnormal ears, clinodactyly, short little fingers
Watson & Gordon (1973)	47, XX +15q-	pter-q22	+	+		+	synophrys, strabismus, hypotonia

TABLE 40 (continued)

Study	Chromosome constitution	Trisomic portion	M.R.	seizures	dermatoglyphic abnormality	bisatellited	Other stigmata
Howard et al. (1974)	47, XX +15q-	pter-q21	+				hyperactive, destructive
Speed et al. (1976)	47, X? +15q-	pter-q22	+			+	
Van Dyke et al. (1976)	47 XY + dic/ 48, XY +dic+dic	p&q prox.*	+	-	-	+	hyperactivity, strabismus, abnormal EEG
Mankinen et al. (1976)	47, XX +15q-	pter-q15	+	-			micrognathia, cleft palate, hooked nose, low set ears, small, slender, irregular dentition, small mouth, clinodactyly, decreased subcutaneous tissue
Pfeiffer & Kessel (1976)	47, XY +15q-	pter-q22	+	+	-	+	hypotonia, pectus excavatum, small
Cohen et al. (1975)	47, XX +der 15 t(15;21)(q22; q22) mat	pter-q22	+	-	-	-	microcephaly, micropthalmia, short neck, nystagmus, dacryostenosis, pilonidal sinus, r. calcaneovalgus, l. metatarsovarus, dysplastic ears, rocker bottom feet, spasticity with contractures

TABLE 40 (continued)

Study	Chromosome constitution	Trisomic portion	M.R.	seizures	dermatoglyphic abnormality	bisatellited	Other stigmata
Bannister & Engel (1975)	47, XY +der 15	p&q prox.	+	-		-	malformed ears, high arched palate, micrognathia, beaked nose, inguinal hernia, micropenis, hypoplastic scrotum, clinodactyly, asymmetric proximal femoral epiphyses, syndactyly of r. second and third toes, syndactyly or l. third and fourth toes
Castel et al. (1976)	46, XX +(7;15)(q35;q14)	pter-q14	+	-	-	-	small, microcephaly, hypertelorism, epicanthal folds, oblique palpebral fissures, long philtrum, camptodactyly
Pfeiffer & Kessel (1976)							
SIBS a.	47, XY +der 15 +(7;15)(q35q14)	pter-q14	+	-	+	-	cleft lip and palate, hypertelorism, strabismus, hyperopia, micrognathia, large ears, pectus excavatum, scoliosis, hypospadias, hypotonia, EEG abnormalities
SIBS b.	47, XY+der 15 +(7;15)(q35;q14)	pter-q14	+	-	+	-	cleft lip and palate, hypertelorism, strabismus, hyperopia, micrognathia, large ears, hypotonia, pectus excavatum
Present study: I	47, XX + mar		+	+	+	+	high arched palate, varus of heel
II	47, XX + mar		+	+	+	+	increased carrying angle, multiple nevi of face and chest

* suggested to be tetrasomic by authors

certain of the translocation trisomy individuals. The remaining ten individuals, however, exhibit only minimal phenotypic abnormalities. The most commonly seen abnormality in this group is seizures, which occur in six of the ten individuals, but is not seen in any of the five individuals with partial trisomy 15 verified through a translocation, or in either of the two individuals with a supernumerary chromosome and multiple abnormalities. This unexpected difference between the two categories of partial trisomy 15 is made even more striking when it is considered that, for three of the five translocation trisomy individuals (Castel et al. and Pfeiffer and Kessel), the trisomic segment is actually smaller than in most of the cases with an extra chromosome.

The clinical data, then, indicate that these two types of partial trisomy 15 are at least two separate abnormalities (i.e., reports of partial trisomy 15 in association with a supernumerary chromosome are probably related to an abnormality other than partial trisomy 15). This idea is supported by cytogenetic evidence; while the occurrence of bisatellited chromosomes is rare and not usually seen in association with mental retardation syndromes, no less than six of the 12 supernumerary chromosomes suspected of being deleted chromosomes 15 display satellites on both chromosome arms. This finding suggests a common, complex origin for these chromosomes and, in fact, in the two instances where centromeric banding was carried out (Van Dyke et al., Pfeiffer and Kessel) the bisatellited chromosomes were found to have two centromeric bands.

Taken together, the clinical and cytogenetic evidence strongly suggest the existence of a distinct chromosome syndrome (different from partial trisomy 15) among those individuals with supernumerary chromosomes listed in Table 40. Several points may be made in support of this idea:

1. the high frequency of bisatellited chromosomes among these individuals; of 14 individuals (12 from previous studies and two from the present study), nine were seen to have an extra, bisatellited chromosome; in all four cases where centromeric banding was performed, the chromosomes were seen to be dicentric (indicating a common mechanism of origin)
2. the relative absence of stigmata among the individuals; only two of the 12 individuals with supernumerary chromosomes displayed multiple physical abnormalities; the remaining individuals, including all individuals with bisatellited chromosomes, appear to have fewer somatic effects
3. the high frequency of seizures; eight of the 14 individuals (and six of the eight with bisatellited chromosome) are known to have had seizures, compared with none of the five translocation trisomy individuals.

Further support for the idea of a distinct chromosome syndrome comes from the study of Newton et al. (1972). In a survey of mentally retarded individuals, two individuals were seen with an extra, bisatellited chromosome midway in size between a D and G group chromosome. Banding of the chromosomes was not done. Both individuals had seizures, but few other obvious abnormalities.

Several authors have speculated on the origin and nature of the bisatellited chromosomes in these individuals. If the assumption is made that most, if not all, of these chromosomes are dicentric in nature, then the following mechanisms could result in such chromosomes: crossing over following a paracentric inversion, asymmetric reciprocal translocation, and sister chromatid union following an isochromatid break (Figure 16). The first and last mechanisms would result in partial tetrasomy for the chromosome involved; the second mechanism could either result in partial tetrasomy (translocation between homologues) or double partial trisomy (translocation between non-homologues).

Van Dyke et al. (1976) have suggested that the bisatellited chromosome seen in their case arose from a sister chromatid union following a break in the long arm of chromosome 15, making the affected individual effectively tetrasomic for the proximal portion of the long arm and all of the short arm of 15. Their case was complicated by the presence of another, smaller, bisatellited chromosome, apparently derived through a bridge-breakage-fusion cycle from the larger bisatellited chromosome. Through an analysis of heteromorphisms, Pfeiffer and Kessel (1976) hypothesized that the bisatellited chromosome in their case was maternal in origin, and resulted from an asymmetrical translocation involving chromosomes 22 and 15. In the present study, an analysis of heteromorphisms in the two individuals revealed differences (in both individuals) between the markers

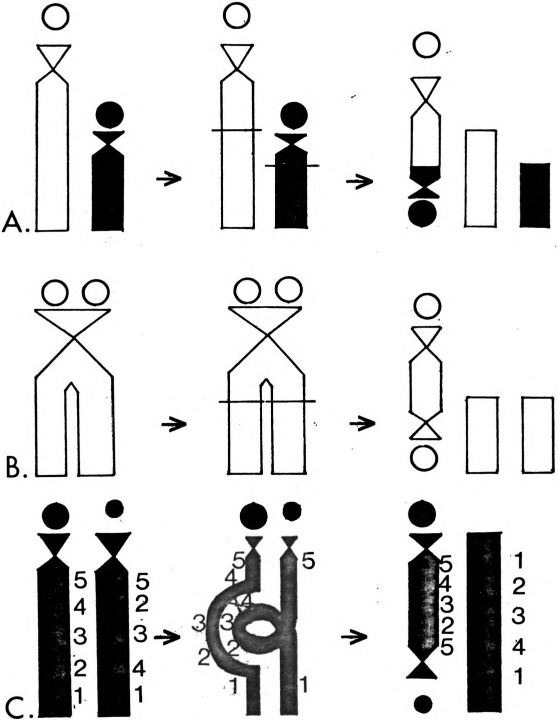

FIGURE 16. Three possible mechanisms leading to the production of a bisatellited, dicentric chromosome: A. asymmetrical reciprocal translocation, B. isochromatid break followed by proximal sister union, C. crossing over following a paracentric inversion

of the two chromosome ends (Figure 11). This would eliminate sister chromatid union following an isochromatid break as a possible mechanism for the origin of these chromosomes. Analysis of C-band heteromorphisms in both individuals revealed that centromeric regions were consistently darker than would be expected if the chromosomes were derived from either chromosome 13 or 21. Further attempts to specify the identity and origin of the bisatellited chromosomes seen in the present study were unsuccessful. We are presently only able to conclude that the bisatellited chromosomes arose from either an asymmetric reciprocal translocation involving chromosomes 14, 15, or 22, or as result of crossing over following a paracentric inversion in any of these three chromosomes.

SUMMARY

Almost all previous dermatoglyphic studies of mental retardation have involved characterizations of abnormalities seen in association with well-defined syndromes. In the present study we have attempted to do the reverse; that is, to determine the usefulness of dermatoglyphic analysis as a screening technique for the detection of new mental retardation syndromes.

Fingerprints were collected from 868 non-specific mentally retarded individuals (537 males, 331 females) and 1463 control individuals (733 males, 730 females). The fingerprints of the retarded and control groups were compared initially with respect to two quantitative traits - total finger ridge count and pattern intensity. A significant decrease in total finger ridge count was seen among the retarded males; however, no other significant differences were seen between the control and retarded groups with respect to these traits. Subsequent analysis of frequency distribution of the four digital patterns showed that, among the retarded males, there was a significant increase in arches and a decrease in whorls; among the females, there was a significant increase in both whorls and arches, and a decrease in ulnar loops.

Further analysis of fingerprint patterns involved an analysis of ten-finger patterns, in an attempt to determine if any unusual

pattern combinations existed in excess among the retarded population. Eighteen such types of ten-finger patterns were isolated, most of which involved the presence of either radial loops or arches (either in high frequency or on digits other than expected).

Forty-seven individuals with either of two of these types of ten-finger patterns (i.e., arches or radial loops on digit I) were subsequently examined for possible similarities indicative of a mental retardation syndrome. Among these individuals, higher than expected frequencies of cryptorchidism (8/28 males) and autosomal chromosome abnormalities (4/47) were noted. Two of the individuals with chromosome abnormalities were found to have a similar cytogenetic abnormality, involving the presence of a supernumary, bisatellited, dicentric chromosome. Clinical and cytogenetic similarities between these two individuals (and others reported in the literature as having partial trisomy 15) are consistent with that of a chromosomal syndrome.

APPENDICES

1 SOURCE OF DATA	2 EXAMINATION DATE	3 HEIGHT (cm)	4 WEIGHT (lbs)	FOR STATE USE ONLY: HT IN PERCENTILE	HT AGE	WT IN PERCENTILE	WT AGE
1 ☐ Chart 2 ☐ Examination							

HEAD	1. Circumference (cm) 1. └─┴─┘ 2. ☐ UK	2 Frontal Bossing 1 ☐ Yes 2 ☐ No 3 ☐ UK	3. Abnormal Head Shape 1 ☐ Yes** 2 ☐ No 3 ☐ UK	4 Anterior Fontanelle 1 ☐ Open 2 ☐ Closed 3 ☐ UK	If open give size in cm Width └─┴─┘ Length └─┴─┘
NECK	5. Webbed Neck, Short Neck 1. ☐ Yes 2 ☐ No 3. ☐ UK	6. Low Posterior Hairline 1 ☐ Yes 2. ☐ No 3 ☐ UK	7 Other**		
FACE & EARS	8. Facial Appearance Abnormal? 1. ☐ Yes** 2 ☐ No 3 ☐ UK	9. Flat Nasal Bridge 1 ☐ Yes 2 ☐ No 3. ☐ UK	10 Misshapen Ears 1 ☐ Yes 2 ☐ No 3 ☐ UK	11 Low Set Ears 1 ☐ Yes 2 ☐ No 3 ☐ UK	12 Other**
EYES	13. Interpupillary Distance (cm) 1 └─┴─┘ 2. ☐ UK	14. Slanted Eyes 1. ☐ Mongoloid 2 ☐ Anti Mongoloid 3. ☐ Normal 4. ☐ UK		15. Strabismus 1. ☐ Yes 2 ☐ No 3 ☐ UK	17. Nystagmus 1 ☐ Yes 2 ☐ No 3 ☐ UK
	18. Retinitus Pigmentosa 1. ☐ Yes 2 ☐ No 3 ☐ UK	19 Epicanthal Fold 1. ☐ Yes 2. ☐ No 3 ☐ UK	20. Macula Cherry Red Spot 1. ☐ Yes 2 ☐ No 3. ☐ UK	21 Ptosis 1. ☐ Yes 2 ☐ No 3 ☐ UK	22. Cataract 1. ☐ Yes 2 ☐ No 3. ☐ UK
	23 Other**	24. Fundoscopy 1. ☐ Done 2 ☐ Not Done 3. ☐ UK	Findings		
MOUTH	25 Cleft Lip 1. ☐ Yes 2 ☐ No 3 ☐ UK	26 Cleft Palate 1 ☐ Yes 2 ☐ No 3 ☐ UK	27. High Arched Palate 1 ☐ Yes 2 ☐ No 3 ☐ UK	28. Malocclusion 1 ☐ Yes 2 ☐ No 3 ☐ UK	29. Discolored Enamel 1. ☐ Yes 2 ☐ No 3. ☐ UK
	30. Enlarged Tongue 1 ☐ Yes 2 ☐ No 3. ☐ UK	31. Micro gnathia 1. ☐ Yes 2 ☐ No 3 ☐ UK	32 Other**		
CHEST	33 Circumference 1 └─┴─┘ 2 ☐ UK	34 Pectus Carinatum 1 ☐ Yes 2 ☐ No 3. ☐ UK	35 Pectus Excavatum 1 ☐ Yes 2 ☐ No 3. ☐ UK	36 Wheeze 1 ☐ Yes 2 ☐ No 3. ☐ UK	37 Other**
HEART	38 Enlarged 1. ☐ Yes 2 ☐ No 3. ☐ UK	39. Murmur 1 ☐ Yes 2. ☐ No 3 ☐ UK	Grade Type		40 Other**
ABDOMEN	40. Enlarged Liver 1 ☐ Yes 2. ☐ No 3. ☐ UK	41. Enlarged Spleen 1. ☐ Yes 2 ☐ No 3. ☐ UK	42. Hernia Umbilical 1 ☐ Yes 2 ☐ No 3 ☐ UK	43 Rectus Diastasis 1. ☐ Yes 2 ☐ No 3 ☐ UK	44. Other**
GENITALIA FEMALE	45. Fused Labia 1 ☐ Yes 2 ☐ No 3. ☐ UK	46. Enlarged Clitoris 1 ☐ Yes 2. ☐ No 3 ☐ UK	47 Vagina Abnormal? 1 ☐ Yes** 2 ☐ No 3. ☐ UK	48 Inguinal Hernia 1 ☐ Yes 2 ☐ No 3. ☐ UK	49 Other**
GENITALIA MALE	50 Micro penis 1. ☐ Yes 2 ☐ No 3. ☐ UK	51 Testes Size (cm) Rt 1 └─┴─┘ Lt. 2 └─┴─┘ 3 ☐ UK	52 Undescended Testes 1 ☐ Rt 2 ☐ Lt 3. ☐ UK	53 Inguinal Hernia 1 ☐ Yes 2 ☐ No 3 ☐ UK	54 Other**
EXTREMITIES	55. Clinodactyly 1 ☐ Hands 2. ☐ Feet 3 ☐ UK	56 Polydactyly 1 ☐ Hands 2 ☐ Feet 3. ☐ UK	57 Syndactyly 1 ☐ Hands 2. ☐ Feet 3 ☐ UK	58 Scoliosis 1 ☐ Yes 2 ☐ No 3. ☐ UK	59. Spina Bifida 1 ☐ Yes 2 ☐ No 3 ☐ UK
	60. Absent Fingers, Toes 1. ☐ Yes 2 ☐ No 3 ☐ UK	61 Overlapping Fingers, Toes 1 ☐ Yes 2 ☐ No 3. ☐ UK	62 Abnormal Fingers, Toes 1 ☐ Yes 2 ☐ No 3 ☐ UK	63 Contracture 1 ☐ Yes 2. ☐ No 3 ☐ UK	64 Simian Line 1. ☐ Yes 2 ☐ No 3. ☐ UK
	65. Joints, Hyper Extendible 1. ☐ Yes 2. ☐ No 3 ☐ UK	66. Other**	67 Span (Length or width in cm) 1 └─┴─┘ 2. ☐ UK		

*See reverse side for codes

**Describe conditions on seperate sheet

APPENDIX A

Physical examination form used in evaluating the majority of individuals with abnormal thumbprints (obtained from Oakdale Developmental Center, Lapeer, Michigan

APPENDIX B

Ten finger patterns in control and mentally retarded males grouped according to the three number codes described in Tables 13 and 14.

For each ten finger pattern:

1 = ulnar loop
2 = whorl
3 = radial loop
4 = arch

n - control = 730
profound - severe = 278
moderate - mild = 259

	Control	Profound - Severe	Moderate - Mild
1-1-1	n	n	n
11111 11111	37	19	13
11112 11111	1	0	0
11111 11121	1	0	1
11112 11122	1	1	0
11114 11124	0	1	0
11121 11111	10	2	5
11122 11111	0	2	1
11121 11121	3	1	1
11121 11122	3	2	0
11122 11121	1	1	0
11122 11122	0	1	0
11121 11131	0	0	1
11131 11111	2	0	0
11132 11111	1	0	0
11131 11131	0	1	0
1-1-2			
11111 11221	0	0	1
11112 11222	0	1	0
11121 11221	0	1	0

	Control	Profound - Severe	Moderate - Mild
1-1-3	n	n	n
11221 11111	2	0	1
11222 11111	1	0	0
11221 11122	0	0	1
11222 11122	0	0	1
1-1-4			
11221 11221	1	0	0
11222 11221	0	0	1
1-1-5			
11111 11331	2	0	0
11111 11412	1	0	0
11141 11411	0	0	1
11411 11111	2	0	0
11411 11114	0	1	0
11121 11421	0	0	1
11211 11311	0	0	1
11421 11411	0	0	1
11421 11422	0	0	1
11443 11411	1	0	0

	Control n	Profound - Severe n	Moderate - Mild n
1-2-1			
11111 12111	1	0	2
11121 12111	5	0	0
11121 12121	0	1	0
11122 12112	1	0	0
11111 12121	0	1	0
11122 12121	1	0	0
11122 12122	1	0	0
1-2-3			
11221 12121	1	0	0
11222 12122	1	0	0
1-2-4			
11221 12221	1	0	0
11221 12222	0	1	0
1-2-5			
11222 12311	0	1	0
11412 12412	1	0	0
11411 12421	0	0	0
1-3-1			
11111 13111	16	4	6
11111 13112	0	1	0
11111 13131	0	1	0
11112 13121	1	1	0
11121 13111	2	0	0
11121 13121	4	1	1
11131 13111	1	0	1
1-3-3			
11211 13111	0	0	1
1-3-5			
11111 13311	1	1	0
11111 13312	0	1	0
11111 13412	0	0	1

	Control n	Profound - Severe n	Moderate - Mild n
1-3-5			
11311 13111	1	0	0
11411 13111	1	0	0
11411 13412	1	0	0
11121 13321	1	0	0
1-4-1			
11111 14111	3	2	1
11111 14141	0	1	0
11121 14111	0	1	0
11121 14121	0	0	1
11121 14122	0	0	1
1-4-5			
11111 14411	1	0	0
11131 14411	1	0	0
11311 14111	0	2	0
11411 14111	0	0	1
11422 14122	0	1	0
1-5-1			
12111 11111	6	4	3
12113 11112	0	1	0
12121 11111	0	0	1
12122 11111	1	1	1
12121 11121	2	0	2
12122 11121	2	1	0
12122 11122	0	1	0
1-5-2			
12121 11212	1	0	0
12121 11221	2	1	2
12122 11211	0	0	1
12122 11221	1	0	0
12122 11222	1	0	1
1-5-3			
12211 11111	0	1	0

	Control	Profound - Severe	Moderate - Mild
1-5-3	n	n	n
12221 11111	0	1	0
12222 11112	1	0	0
12222 11121	0	1	0
1-5-4			
12211 11211	0	1	0
12211 11221	0	1	0
12222 11211	0	1	0
12222 11221	0	0	1
1-5-5			
12211 11311	0	0	2
1-6-1			
12111 12111	2	1	2
12111 12121	0	1	0
12112 12122	0	0	1
12121 12111	1	2	0
12121 12121	3	0	0
12122 12111	0	1	0
12122 12121	1	0	0
12122 12112	0	2	0
12122 12122	1	0	0
12131 12111	0	0	1
1-6-2			
12111 12221	1	0	0
12121 12211	0	1	0
12121 12221	3	0	1
12122 12222	1	1	0
1-6-3			
12211 12111	1	0	0
12221 12111	2	0	0
12221 12121	2	1	0
12222 12121	3	0	0
12222 12122	1	0	1

	Control	Profound - Severe	Moderate - Mild
1-6-4	n	n	n
12221 12211	1	0	0
12221 12221	2	3	0
12221 12221	2	3	0
12222 12221	2	1	2
12222 12222	2	2	0
1-6-5			
12111 12411	1	0	0
12421 12111	1	0	0
1-7-1			
12111 13111	1	0	1
12111 13121	1	0	0
12121 13111	2	0	0
12121 13122	1	0	0
12121 13121	0	1	0
1-7-2			
12121 13221	0	1	0
1-7-3			
12221 13111	1	0	0
1-7-4			
12221 13221	1	0	1
12222 13222	1	0	0
1-7-5			
12111 13322	1	0	0
12121 13321	0	0	1
1-8-2			
12112 14221	1	0	0
12121 14221	0	1	0

1-9-1	Control n	Profound - Severe n	Moderate - Mild n
13111 11111	17	1	7
13111 11121	3	0	0
13121 11111	8	1	1
13121 11121	1	0	1
13122 11121	1	0	0
13131 11111	0	0	1

1-9-2	Control n	Profound - Severe n	Moderate - Mild n
13111 11211	1	0	0

1-9-3	Control n	Profound - Severe n	Moderate - Mild n
13211 11111	1	0	0
13221 11121	1	0	1
13222 11122	1	0	0

1-9-5	Control n	Profound - Severe n	Moderate - Mild n
13111 11311	1	1	0
13311 11311	1	0	0
13311 11411	0	0	1
13411 11111	1	0	0
13411 11311	1	0	0
13321 11111	1	1	0
13421 11111	0	1	0

1-10-1	Control n	Profound - Severe n	Moderate - Mild n
13111 12111	2	1	2
13121 12111	1	1	1
13121 12121	1	1	0
13122 12111	0	1	0
13122 12122	0	1	0

1-10-2	Control n	Profound - Severe n	Moderate - Mild n
13111 12221	1	0	0

1-10-3	Control n	Profound - Severe n	Moderate - Mild n
13211 12111	0	1	0
13221 12111	1	0	0
13221 12121	1	0	0
13231 12111	1	0	0

1-10-4	Control n	Profound - Severe n	Moderate - Mild n
13221 12221	1	0	1
13222 12221	1	0	0

1-10-5	Control n	Profound - Severe n	Moderate - Mild n
13411 12411	0	0	1

1-11-1	Control n	Profound - Severe n	Moderate - Mild n
13111 13111	22	7	9
13111 13121	3	0	0
13121 13111	3	2	2
13121 13121	3	1	0
13122 13121	1	0	0

1-11-3	Control n	Profound - Severe n	Moderate - Mild n
13211 13111	1	0	0
13221 13111	0	0	1

1-11-4	Control n	Profound - Severe n	Moderate - Mild n
13211 13211	1	0	0
13211 13221	0	1	0
13221 13221	0	0	1

1-11-5	Control n	Profound - Severe n	Moderate - Mild n
13111 13311	1	0	0
13311 13111	3	0	4
13111 13411	0	0	1
13321 13311	0	0	1
13411 13111	3	1	0
13411 13411	2	0	0

1-12-1	Control n	Profound - Severe n	Moderate - Mild n
13111 14111	1	0	1

	Control n	Profound - Severe n	Moderate - Mild n
1-12-1			
13121 14111	0	0	1
1-12-5			
13111 14411	2	0	1
13411 14441	0	1	0
1-13-1			
14111 11111	4	1	5
14121 11111	1	1	0
14121 11121	1	0	0
14131 11111	1	0	0
1-13-5			
14111 11311	0	1	0
14411 11411	1	0	0
14121 11411	1	0	1
14131 11441	0	1	0
14422 11222	0	0	1
14441 11411	1	0	0
1-14-1			
14111 12111	1	0	1
14111 12121	0	1	0
14122 12122	0	0	1
1-15-1			
14111 13111	5	1	2
14111 13121	1	0	0
14121 13111	1	0	0
14121 13121	2	1	0
1-15-5			
14121 13121	1	0	0
14111 13341	1	0	0
14112 13422	1	0	0
14121 13411	1	0	0
14144 13441	0	1	0

	Control n	Profound - Severe n	Moderate - Mild n
1-15-5			
14311 13111	0	0	1
14411 13311	1	0	0
14411 13411	1	1	0
14411 13441	1	0	0
1-16-1			
14111 14111	5	2	4
14111 14121	0	0	1
14111 14141	1	0	0
14121 14111	1	1	0
14124 14111	1	0	0
14121 14121	1	0	0
14121 14124	1	0	0
14131 14121	1	0	0
1-16-5			
14111 14411	5	1	2
14111 14444	0	0	2
14112 14411	0	1	0
14121 14411	1	0	0
14144 14414	1	0	0
14321 14111	0	1	0
14411 14111	1	0	0
14411 14411	1	1	1
14414 14114	0	1	0
14414 14411	0	0	1
14414 14444	0	1	0
14441 14412	0	0	1
14444 14414	1	0	0
14441 14441	1	0	0
14444 14444	0	1	0
2-1-1			
11111 21111	0	1	0
11121 21121	0	1	0
2-1-2			
11121 21222	1	0	0

2-1-5	Control n	Profound - Severe n	Moderate - Mild n
11111 21311	0	1	0
11111 21321	0	1	0
11123 21421	0	1	0
11131 21421	1	0	0
2-2-1			
11111 22111	1	0	1
11121 22111	0	0	1
2-3-1			
11111 23111	4	0	0
11111 23121	0	0	1
11122 23122	1	0	0
2-3-5			
11111 23411	1	0	0
11411 23121	1	0	0
2-4-1			
11111 24142	1	0	0
2-4-5			
11411 24111	0	0	1
2-5-1			
12111 21111	2	0	0
12111 21121	1	0	0
12121 21122	1	0	0
12122 21122	1	0	0
2-5-2			
12121 21222	1	0	0
2-5-4			
12221 21221	2	0	0

2-6-1	Control n	Profound - Severe n	Moderate - Mild n
12111 22111	4	0	1
12121 22121	1	0	0
12121 22122	1	0	0
12122 22121	0	0	1
2-6-2			
12111 22211	1	0	0
12121 22221	0	1	0
12122 22221	0	0	1
2-6-4			
12221 22211	1	0	0
12221 22221	1	0	1
12222 22221	1	0	0
2-6-5			
12122 22432	1	0	0
2-7-1			
12111 23111	0	1	0
2-7-2			
12121 23221	0	0	2
2-9-1			
13111 21111	1	0	1
13121 21121	1	0	0
13121 21122	0	0	1
2-9-5			
13111 21311	1	0	0
2-10-3			
13221 22121	0	1	0

	Control	Profound - Severe	Moderate - Mild
2-10-4	n	n	n
13221 12222	0	1	0
2-11-1			
13111 23111	3	0	0
13111 23121	2	0	0
2-11-2			
13111 23221	1	0	0
2-14-3			
14222 22122	0	1	0
2-15-1			
14111 23111	0	0	1
2-16-5			
14114 24414	2	0	0
14411 24411	1	0	0
3-1-1			
21111 11111	12	3	4
21121 11111	4	1	0
21121 11121	2	0	2
21122 11111	1	0	0
21122 11121	1	0	0
21123 11111	0	1	0
3-1-2			
21122 11222	1	0	0
3-1-3			
21222 11111	2	0	0
3-1-5			
21111 11411	0	1	0

	Control	Profound - Severe	Moderate - Mild
3-1-5	n	n	n
21421 11221	1	0	0
21422 11421	0	1	0
3-2-1			
21111 12111	0	0	1
21112 12122	1	0	0
21121 12111	1	0	0
21121 12121	1	0	0
21122 12122	2	0	0
3-3-1			
21111 13111	5	0	0
3-3-2			
21121 13221	0	1	0
3-3-4			
21222 13221	0	1	0
3-3-5			
21111 13311	0	1	0
21111 13411	0	0	1
21321 13111	1	0	0
3-4-1			
21111 14111	2	0	1
3-5-1			
22111 11111	4	0	1
22111 11121	1	0	0
22121 11111	1	0	1
22121 11122	0	0	1
22121 11121	6	1	0
22122 11111	1	0	0
22122 11122	2	1	0

	Control	Profound - Severe	Moderate - Mild
<u>3-5-2</u>	<u>n</u>	<u>n</u>	<u>n</u>
22121 11222	1	0	0
22121 11222	0	1	0
<u>3-5-3</u>			
22221 11121	0	1	0
22222 11122	0	0	1
<u>3-5-4</u>			
22222 11222	0	0	1
<u>3-5-5</u>			
22111 11311	0	1	0
<u>3-6-1</u>			
22111 12111	6	0	0
22112 12111	1	0	0
22112 12112	0	1	0
22111 12121	1	0	0
22121 12111	4	0	0
22121 12121	3	0	2
22122 12121	2	0	0
22122 12122	1	0	0
<u>3-6-2</u>			
22111 12211	1	0	0
22121 12211	1	0	0
<u>3-6-3</u>			
22211 12121	0	0	1
22221 12111	3	1	0
22222 12121	0	1	0
22222 12122	1	0	0
<u>3-6-4</u>			
22211 12221	1	0	0
22221 12221	1	0	2

	Control	Profound - Severe	Moderate - Mild
<u>3-6-4</u>	<u>n</u>	<u>n</u>	<u>n</u>
22222 12211	0	1	0
22221 12222	1	0	0
22222 12221	6	2	0
22222 12222	6	2	0
<u>3-7-1</u>			
22111 13111	2	0	0
<u>3-7-2</u>			
22111 13221	1	0	0
<u>3-7-5</u>			
22421 13122	1	0	0
<u>3-8-1</u>			
22121 14121	1	0	0
<u>3-9-1</u>			
23111 11111	0	1	0
23121 11111	3	1	0
23131 11111	0	0	1
<u>3-9-3</u>			
23221 11111	1	1	0
23221 11121	1	0	0
<u>3-9-4</u>			
23221 11221	0	1	0
<u>3-9-5</u>			
23321 11111	2	0	0
23411 11411	0	0	1
<u>3-10-1</u>			
23111 12111	6	1	0

	Control	Profound - Severe	Moderate - Mild
3-10-1	n	n	n
23121 12111	2	0	1
3-10-3			
23221 12111	0	0	2
23221 12121	1	0	0
23222 12121	1	0	1
23222 12111	0	1	0
3-10-4			
23221 12221	1	0	0
3-10-5			
23322 12111	0	1	0
23311 12411	0	0	1
3-11-1			
23111 13111	9	3	2
23121 13121	0	1	0
23121 13111	0	1	1
3-11-3			
23221 13121	1	0	0
3-11-5			
23111 13311	1	0	0
3-12-1			
23121 14111	1	0	0
23122 14122	0	0	1
3-12-3			
23222 14121	1	0	0
3-12-5			
23111 14421	2	0	0

	Control	Profound - Severe	Moderate - Mild
3-12-5	n	n	n
23421 14411	0	1	0
3-13-1			
24111 11111	4	1	0
3-13-5			
24131 11411	1	0	0
24311 11411	1	0	0
24422 11121	0	0	1
24411 11411	0	0	1
3-15-1			
24111 13111	0	0	1
24121 13111	1	0	0
24121 13122	0	1	0
24122 13111	1	0	0
3-15-5			
24111 13311	0	1	0
24122 13322	0	1	0
3-16-1			
24111 14111	1	0	0
24111 14121	0	1	0
24121 14111	3	0	0
24122 14121	2	0	0
3-16-5			
24411 14111	2	0	0
24411 14411	1	0	0
24414 14411	1	0	0
4-1-1			
21111 21111	11	3	1
21111 21121	2	0	0
21111 21141	1	0	0
21121 21111	4	0	1
21121 21121	3	0	0

4-1-1

	Control n	Profound - Severe n	Moderate - Mild n
21122 21121	2	0	0
21122 21122	1	0	0
21121 21122	0	1	0
21122 21111	0	1	0

4-1-3

	Control n	Profound - Severe n	Moderate - Mild n
21222 21121	1	0	0

4-1-4

	Control n	Profound - Severe n	Moderate - Mild n
21221 21221	0	1	0

4-1-5

	Control n	Profound - Severe n	Moderate - Mild n
21111 21311	0	0	1
21111 21411	1	0	0
21111 21421	1	0	0
21411 21411	1	0	0

4-2-1

	Control n	Profound - Severe n	Moderate - Mild n
21111 22111	0	0	1
21121 22121	2	0	1
21122 22122	0	1	1

4-2-3

	Control n	Profound - Severe n	Moderate - Mild n
21111 22222	1	0	0
21122 22222	1	0	0

4-2-4

	Control n	Profound - Severe n	Moderate - Mild n
21221 22221	1	0	0

4-3-1

	Control n	Profound - Severe n	Moderate - Mild n
21111 23111	1	2	0
21111 23121	0	0	1
21121 23111	1	0	1
21121 23121	0	1	0
21121 23122	1	0	0

4-3-2

	Control n	Profound - Severe n	Moderate - Mild n
21122 23222	0	0	1

4-3-5

	Control n	Profound - Severe n	Moderate - Mild n
21111 23311	0	2	0

4-4-1

	Control n	Profound - Severe n	Moderate - Mild n
21121 24121	0	0	1

4-4-5

	Control n	Profound - Severe n	Moderate - Mild n
21111 24411	0	1	0

4-5-1

	Control n	Profound - Severe n	Moderate - Mild n
22111 21111	2	1	1
22112 21112	1	0	0
22112 21111	0	1	0
22112 21121	1	0	0
22121 21111	2	2	1
22121 21121	3	1	0
22122 21111	0	1	0
22122 21112	1	0	0
22122 21121	1	1	2
22122 21122	1	1	2

4-5-2

	Control n	Profound - Severe n	Moderate - Mild n
22122 21222	0	0	1

4-5-3

	Control n	Profound - Severe n	Moderate - Mild n
22211 21111	1	0	0
22221 21121	0	1	0

4-5-4

	Control n	Profound - Severe n	Moderate - Mild n
22221 21211	1	0	0
22221 21221	1	0	0
22221 21222	1	0	0
22222 21212	0	0	1
22222 21222	4	0	0

4-6-1	Control n	Profound - Severe n	Moderate - Mild n
22111 22111	2	1	0
22111 22112	1	0	0
22111 22121	0	0	1
22112 22111	1	0	0
22121 22111	3	4	2
22121 22112	1	0	0
22121 22121	5	1	0
22121 22122	2	1	1
22122 22111	1	0	0
22122 22112	1	0	0
22122 22121	5	1	0
22122 22122	8	1	0
4-6-2			
22111 22211	1	0	2
22121 22211	1	0	0
22121 22221	2	1	2
22121 22222	1	0	0
22122 22212	1	0	0
22122 22221	3	0	0
22122 22222	3	1	1
4-6-3			
22211 22111	1	1	2
22221 22111	3	0	0
22221 22121	1	0	0
22221 22122	1	1	0
22222 22111	1	1	0
22222 22121	3	0	0
22222 22122	3	0	1
4-6-4			
22211 22212	0	0	1
22221 22211	1	0	0
22222 22211	0	1	0
22221 22221	9	7	1
22221 22222	2	2	0
22222 22221	8	2	4
22222 22222	16	5	7
22222 22224	0	0	1

4-6-5	Control n	Profound - Severe n	Moderate - Mild n
22111 22311	1	1	0
22321 22111	0	0	1
22122 22412	1	0	0
22122 22422	2	0	0
4-7-1			
22111 23111	1	0	0
22111 23121	0	0	1
22121 23111	1	0	0
22121 23122	1	0	0
22122 23121	1	0	0
22122 23122	1	0	0
4-7-2			
22122 23222	1	0	0
4-7-4			
22221 23221	0	0	2
4-7-5			
22122 23311	1	0	0
4-9-1			
23111 21111	3	0	1
23112 21111	1	0	0
23121 21111	3	1	0
23121 21121	2	0	1
23122 21122	0	1	0
4-9-3			
23211 21111	0	1	0
23222 21121	1	0	0
4-9-4			
23222 21222	1	0	0

	Control	Profound - Severe	Moderate - Mild
4-9-5	n	n	n
23121 21411	0	0	1
23311 21111	1	0	0
23411 21111	1	0	0
4-10-1			
23111 22111	2	0	0
23121 22111	2	1	0
23121 22121	1	0	0
23122 22111	1	0	0
23123 22121	0	1	0
4-10-2			
23121 22211	0	1	0
23122 22221	1	0	0
4-10-3			
23211 22111	1	0	0
23221 22111	2	1	0
23222 22121	0	1	0
23222 22122	1	0	0
4-10-4			
23221 22211	2	0	0
23221 22221	1	1	0
4-10-5			
23121 22411	1	0	0
23321 22211	0	0	1
4-11-1			
23111 23111	6	0	2
23121 23111	1	0	0
4-11-2			
23121 23221	0	1	0
23122 23222	1	0	0
4-11-4	n	n	n
23211 23211	0	0	1
4-11-5			
23111 23311	1	0	0
23321 23211	0	0	1
4-12-1			
23111 24111	2	0	0
4-12-5			
23111 24411	2	0	0
4-13-1			
24111 21111	0	0	1
4-13-3			
24211 21111	0	0	1
4-13-5			
24111 21311	0	1	0
24422 21421	0	0	1
4-16-1			
24111 24111	0	0	1
4-16-5			
24421 24111	1	0	0
24411 24411	1	0	1

OTHER - GROUP 5

	Control	Profount - Severe	Moderate - Mild
(3)			
13221 32221	0	0	1
(4)			
11111 41111	0	1	0
11111 41222	0	1	0
11111 41411	0	1	0
12121 41111	0	1	0
12221 42111	0	1	0
12222 43222	0	1	0
13311 43111	0	1	0
14111 44111	0	1	1
14411 44411	0	1	0
14441 44441	0	1	0
11111 44411	0	1	0
11111 44114	0	0	1
11111 43411	0	1	0
14121 44122	0	0	1
14111 44221	0	0	1
11122 43121	1	0	0
11411 44111	1	0	0
12121 42111	1	0	0
12121 43122	1	0	0
13111 41111	1	0	0
13114 41111	1	0	0
14121 43111	1	0	0
14141 43411	1	0	0
14121 41111	1	0	0
(7)			
21111 34411	0	0	1
24431 34411	1	0	0
(8)			
21111 41111	0	0	1
24121 44141	0	1	0
22121 43221	1	0	0
22221 43221	1	0	0

	Control	Profound - Severe	Moderate - Mild
(9)			
34111 14411	0	1	0
(10)			
32222 22222	0	1	0
(11)			
34414 34411	0	0	1
(13)			
34111 13111	0	0	1
43111 14411	1	0	0
41421 12221	0	1	0
43411 14411	0	1	0
44121 11121	0	2	0
44421 14111	0	1	0
(14)			
41111 21111	0	0	1
(16)			
41111 41111	1	0	0
41112 41111	1	0	0
41122 41121	1	0	0
44111 44114	2	0	0
44411 44411	2	0	0
44444 44444	3	2	1
41121 41111	0	1	0
41141 43444	0	1	0
44111 41441	0	1	0
44111 44144	0	1	0
44411 44111	0	1	0
44444 44414	0	1	0
44444 44441	0	1	0
44414 44444	0	0	1
44444 44142	0	0	1
44441 44444	0	0	1

(16)	Control	Profound - Severe	Moderate - Mild
44441 44444	0	0	1
41114 44121	0	0	1
43122 44121	0	0	1
41414 44141	0	1	0

APPENDIX C

Ten finger patterns in control and mentally retarded females grouped according to the three number codes described in Tables 13 and 14.

For each ten finger pattern:

1 = ulnar loop
2 = whorl
3 = radial loop
4 = arch

n - control = 730
profound - severe = 172
moderate - mild = 159

	Control	Profound - Severe	Moderate - Mild
<u>1-1-1</u>	<u>n</u>	<u>n</u>	<u>n</u>
11111 11111	65	13	8
11112 11111	2	0	0
11111 11114	1	0	0
11111 11121	6	0	0
11112 11122	0	0	1
11111 11131	2	0	0
11121 11111	9	5	0
11121 11112	1	0	0
11122 11111	2	0	0
11122 11112	1	0	0
11121 11121	11	0	1
11121 11122	1	0	0
11122 11121	0	0	1
11122 11122	0	0	1
11131 11111	1	1	0
11133 11113	2	0	0
11131 11124	1	0	0
<u>1-1-2</u>			
11111 11211	2	0	0
11111 11221	2	0	1

	Control	Profound - Severe	Moderate - Mild
<u>1-1-2</u>	<u>n</u>	<u>n</u>	<u>n</u>
11121 11221	2	0	0
<u>1-1-3</u>			
11221 11111	3	0	0
11222 11111	1	0	0
<u>1-1-4</u>			
11222 11221	1	0	0
<u>1-1-5</u>			
11111 11311	3	0	0
11111 11411	4	0	0
11111 11422	0	1	0
11121 11421	1	0	0
11441 11411	0	1	0
11441 11444	2	0	0
<u>1-2-1</u>			
11111 12111	7	1	0

	Control	Profound - Severe	Moderate - Mild
1-2-1	n	n	n
11111 12121	2	0	0
11111 12122	1	0	0
11112 12122	2	0	1
11121 12111	1	0	2
11122 12111	2	0	0
11121 12121	4	0	0
11121 12124	0	1	0
11122 12121	0	1	0
11122 12121	1	0	0
1-2-2			
11111 12221	0	0	1
1-2-3			
11221 12121	0	0	1
1-2-4			
11211 12221	1	0	0
1-3-1			
11111 13111	29	6	2
11112 13111	1	0	0
11111 13121	1	0	2
11111 13122	0	0	1
11111 13141	0	0	2
11121 13111	1	1	0
11121 13121	5	1	1
11121 13122	1	0	0
11131 13111	1	0	0
1-3-2			
11111 13221	1	1	0
1-3-5			
11111 13311	4	0	1
11111 13411	3	2	0
11121 13421	1	0	0

	Control	Profound - Severe	Moderate - Mild
1-3-5	n	n	n
11411 13111	0	0	1
11411 13411	2	1	0
1-4-1			
11111 14111	5	1	1
11111 14121	1	0	0
11111 14131	2	0	0
11121 14111	5	0	1
11122 14111	1	0	0
11121 14121	1	0	0
1-4-3			
11122 14111	0	1	0
1-4-5			
11111 14311	1	0	0
11111 14411	2	1	0
1-5-1			
12111 11111	7	0	2
12111 11121	0	1	2
12121 11111	3	0	2
12122 11111	1	0	0
12121 11121	2	0	0
12122 11122	0	0	1
12131 11111	0	1	0
12141 11141	0	1	0
1-5-2			
12121 11211	1	0	0
12121 11221	1	0	0
1-5-3			
12221 11121	5	0	0
1-5-4			
12221 11221	1	0	0

	Control	Profound - Severe	Moderate - Mild
1-5-4	n	n	n
12222 11221	1	0	0
1-5-5			
12321 11111	1	0	0
1-6-1			
12111 12111	15	1	4
12112 12111	2	0	0
12111 12121	1	0	0
12121 12111	5	0	0
12121 12112	1	0	0
12121 12121	3	0	0
12122 12121	2	0	2
12122 12122	0	1	0
1-6-2			
12111 12221	1	0	0
12121 12221	3	1	0
12121 12222	1	0	0
12122 12221	1	0	0
12122 12222	2	0	0
1-6-3			
12211 12121	2	0	0
12221 12111	1	0	0
12222 12122	2	0	1
1-6-4			
12211 12211	1	0	0
12221 12211	0	0	1
12222 12211	0	0	1
12221 12221	4	3	0
12222 12221	1	0	0
12222 12222	1	1	0
1-6-5			
12111 12311	1	0	1

	Control	Profound - Severe	Moderate - Mild
1-6-5	n	n	n
12111 12311	1	0	1
12121 12321	2	0	0
1-7-1			
12111 13111	3	1	1
12121 13111	1	1	0
1-7-2			
12111 13211	2	0	0
12121 13211	2	0	0
12121 13221	1	0	0
1-7-3			
12221 13111	0	1	0
1-7-4			
12221 13221	3	0	0
1-7-5			
12321 13421	0	1	0
1-8-1			
12121 14111	1	0	0
1-9-1			
13111 11111	12	1	3
13111 11112	1	0	0
13111 11121	1	0	0
13121 11111	4	0	0
1-9-2			
13121 11221	0	0	1
1-9-3			
13211 11111	1	0	0

	Control n	Profound - Severe n	Moderate - Mild n
1-9-3			
13221 11111	1	0	0
1-9-5			
13111 11411	2	0	0
13314 11441	0	0	1
1-10-1			
13111 12111	9	1	1
13111 12121	1	0	0
13121 12121	0	1	0
1-10-5			
13111 12321	1	0	0
13111 12411	2	0	0
1-11-1			
13111 13111	27	9	7
1-11-2			
13121 13221	2	0	0
1-11-3			
13211 13111	1	0	0
1-11-5			
13111 13311	3	0	0
13111 13331	0	0	1
13111 13411	2	0	0
13121 13411	2	0	0
13121 13421	1	0	0
13122 13322	0	1	0
13411 13111	2	0	0
1-12-1			
13111 14111	2	0	0
13111 14121	1	0	0
1-12-5			
13111 14411	0	0	1
13411 14411	2	0	0
13441 14441	0	0	1
1-13-1			
14111 11111	6	0	2
14112 11111	0	0	1
14121 11111	1	0	0
14124 11114	0	0	1
1-13-5			
14111 11411	3	2	0
14111 11441	2	0	0
14121 11421	2	0	0
14141 11431	1	0	0
14441 11441	1	0	0
1-14-1			
14111 12121	1	0	0
14121 12121	0	1	0
1-15-1			
14111 13111	3	0	0
14111 13114	0	0	1
14111 13121	2	0	0
14112 13122	0	1	0
14121 13111	1	0	0
1-15-5			
14111 13311	1	0	0
14111 13411	5	0	0
14311 13111	2	0	0
14411 13111	1	0	0
14422 13122	0	0	1
14444 13444	0	1	0
1-16-1			
14111 14111	2	0	2

	Control	Profound - Severe	Moderate - Mild
1-16-1	n	n	n
14111 14121	2	0	0
14111 14121	1	0	0
14121 14114	1	0	0
14121 14121	1	0	0
14122 14121	1	0	0
14131 14114	2	0	0
14144 14144	1	0	0
1-16-5			
14111 14311	2	0	0
14111 14411	2	0	0
14111 14441	2	0	0
14124 14414	1	0	0
14141 14444	0	1	0
14141 14441	1	0	0
14421 14111	2	0	0
14411 14411	5	0	0
14411 14441	1	0	0
14441 14411	0	1	0
14441 14441	6	0	1
14444 14444	1	0	2
2-1-1			
11111 21111	2	1	3
11111 21121	1	0	0
11121 21111	0	0	1
11121 21121	1	0	1
11122 21122	0	1	0
11132 21131	0	0	1
2-1-2			
11111 21211	0	1	0
11121 21221	0	0	1
2-1-5			
11111 21311	1	0	0
2-3-1			
11111 23111	1	0	0

	Control	Profound - Severe	Moderate - Mild
2-3-1	n	n	n
11111 23121	0	0	1
2-3-2			
11121 23222	0	1	0
2-3-5			
11111 23411	1	0	0
11111 23421	1	0	0
2-4-1			
11111 24111	2	0	0
11121 24121	1	0	0
2-5-1			
12111 21111	1	0	0
2-5-2			
12122 21222	1	0	0
2-5-3			
12221 21111	0	1	0
2-5-5			
12111 21311	1	0	0
2-6-1			
12111 22111	3	1	1
12121 22111	1	0	0
12121 22121	1	1	0
12121 22122	1	0	0
2-6-2			
12111 22221	0	1	0
12121 22222	0	1	0

	Control n	Profound - Severe n	Moderate - Mild n
2-6-4			
12221 22211	1	0	0
12221 22222	1	1	0
12222 22222	0	1	1
2-7-1			
12111 23111	0	2	1
12111 23112	0	1	0
12121 23121	0	1	0
2-9-1			
13111 21111	3	0	0
13111 21112	1	0	0
13121 21122	0	1	0
2-10-4			
13211 22221	1	0	0
2-10-5			
13321 22111	0	0	1
2-11-1			
13111 23111	4	0	0
13121 23111	0	0	1
2-11-5			
13111 23311	0	0	1
2-13-1			
14111 21141	0	0	1
2-13-5			
14421 21422	0	1	0
2-16-5			
14111 24411	1	0	0

	Control n	Profound - Severe n	Moderate - Mild n
2-16-5			
14111 24444	1	0	0
14441 24411	0	1	0
3-1-1			
21111 11111	10	2	1
21121 11111	1	1	1
21122 11111	1	0	0
21121 11121	0	1	0
3-1-2			
21111 11211	0	1	0
21122 11222	1	0	0
3-1-5			
21411 11111	1	0	0
21411 11411	0	1	0
3-2-1			
21111 12111	1	0	1
21121 12121	0	1	0
21122 12121	1	0	0
3-2-2			
21111 12221	0	1	0
3-2-4			
21222 12221	1	0	0
3-3-1			
21111 13111	1	0	0
21111 13121	1	0	0
21121 13122	1	0	0
3-3-4			
21221 13211	1	0	0

3-3-5	Control n	Profound - Severe n	Moderate - Mild n
21124 13424	1	0	0

3-4-1	Control n	Profound - Severe n	Moderate - Mild n
21111 14111	1	0	0

3-5-1	Control n	Profound - Severe n	Moderate - Mild n
22111 11111	1	0	2
22111 11141	0	1	0
22121 11111	2	0	1
22121 11121	1	0	0

3-5-3	Control n	Profound - Severe n	Moderate - Mild n
22211 11111	1	0	0
22221 11111	2	0	0
22222 11122	0	0	1

3-5-4	Control n	Profound - Severe n	Moderate - Mild n
22211 11211	0	0	1
22221 11221	1	0	0

3-5-5	Control n	Profound - Severe n	Moderate - Mild n
22111 11411	1	0	0

3-6-1	Control n	Profound - Severe n	Moderate - Mild n
22111 12111	5	0	1
22112 12111	0	1	0
22111 12121	0	0	1
22111 12122	0	1	0
22121 12111	0	0	1
22122 12111	0	0	1
22121 12121	0	1	0
22121 12122	0	1	0
22122 12122	0	0	1

3-6-2	Control n	Profound - Severe n	Moderate - Mild n
22111 12221	0	1	0
22121 12221	2	0	0
22122 12222	1	0	2

3-6-3	Control n	Profound - Severe n	Moderate - Mild n
22211 12111	0	0	1
22221 12121	1	0	0
22222 12122	1	0	0

3-6-4	Control n	Profound - Severe n	Moderate - Mild n
22221 12211	1	0	0
22221 12221	2	0	0
22221 12222	1	0	0
22222 12221	1	1	1
22222 12222	0	0	1

3-6-5	Control n	Profound - Severe n	Moderate - Mild n
22111 12411	1	0	0

3-7-1	Control n	Profound - Severe n	Moderate - Mild n
22111 13111	0	1	0
22122 13122	1	0	0

3-7-4	Control n	Profound - Severe n	Moderate - Mild n
22211 13221	1	0	0
22222 13211	0	0	1

3-7-5	Control n	Profound - Severe n	Moderate - Mild n
22111 13111	1	0	0
22121 13321	0	2	0

3-9-1	Control n	Profound - Severe n	Moderate - Mild n
23111 11111	0	1	0
23121 11111	0	1	0
23131 11121	1	0	0

	Control	Profound - Severe	Moderate - Mild
<u>3-9-3</u>	<u>n</u>	<u>n</u>	<u>n</u>
23221 11121	1	0	0
<u>3-10-1</u>			
23111 12111	4	0	0
<u>3-10-2</u>			
23122 12221	0	1	0
<u>3-10-3</u>			
23222 12111	0	0	2
<u>3-11-1</u>			
23111 13111	1	1	0
23121 13111	0	1	0
23121 13121	0	0	1
23121 13122	1	0	0
<u>3-11-5</u>			
23111 13311	1	0	0
23121 13421	1	0	1
<u>3-13-1</u>			
24111 11111	1	0	0
24121 11111	2	0	0
<u>3-15-1</u>			
24121 13111	1	0	0
<u>3-15-5</u>			
24411 13411	1	0	0
<u>3-16-1</u>			
24111 14111	1	0	0
24111 14114	0	1	0

	Control	Profound - Severe	Moderate - Mild
<u>3-16-5</u>	<u>n</u>	<u>n</u>	<u>n</u>
24311 14121	0	1	0
24411 14411	1	0	0
<u>4-1-1</u>			
21111 21111	4	0	3
21111 21121	1	1	1
21121 21111	5	0	0
21122 21111	0	1	0
21121 21121	4	2	0
21121 21122	1	0	0
21122 21121	1	0	0
21122 21122	1	0	0
<u>4-1-4</u>			
21221 21221	1	0	0
<u>4-1-5</u>			
21111 21311	1	0	0
21411 21111	1	0	0
<u>4-2-1</u>			
21111 22111	1	0	0
21121 22111	1	1	1
21122 22121	0	0	1
21122 22122	2	0	0
21122 22142	1	0	0
<u>4-2-2</u>			
21111 22211	1	0	0
21121 22221	0	0	1
<u>4-2-3</u>			
21221 22121	0	0	1
<u>4-2-4</u>			
21221 22222	1	0	0

	Control	Profound - Severe	Moderate - Mild
4-2-5	n	n	n
21111 22411	1	0	0
4-3-1			
21111 23111	1	0	0
21121 23121	1	0	0
21122 23122	1	0	0
4-3-5			
21111 23411	1	0	0
21111 23421	1	0	0
4-4-1			
12111 24111	1	0	0
4-4-5			
21122 24421	1	0	0
4-5-1			
22111 21111	2	1	1
22111 21122	0	1	0
22122 21122	2	0	0
4-5-2			
22122 21221	1	0	0
4-5-4			
22221 21221	2	1	0
4-6-1			
22111 22111	3	2	0
22111 22112	0	0	1
22111 22121	1	0	0
22121 22111	1	1	0
22121 22121	3	0	3
22121 22122	1	0	0

	Control	Profound - Severe	Moderate - Mild
4-6-1	n	n	n
22122 22121	0	0	1
22122 22122	5	0	2
4-6-2			
22112 22212	1	0	0
22111 22221	2	0	0
22112 22222	1	0	0
22121 22221	2	0	0
22121 22222	3	1	0
22122 22221	2	0	0
22122 22222	2	0	1
4-6-3			
22221 22111	1	2	0
22222 22111	2	0	0
22221 22121	2	1	0
22221 22122	1	0	0
22222 22121	1	0	1
22222 22122	1	0	1
4-6-4			
22211 22221	2	0	0
22221 22211	0	0	1
22222 22212	1	0	0
22221 22221	15	3	1
22221 22222	1	1	0
22222 22222	9	8	4
4-6-5			
22121 22311	0	1	0
22121 22321	0	1	0
22121 22322	0	1	0
22222 22322	1	0	0
4-7-1			
22111 23111	4	0	0
22111 23121	1	0	0
22121 23111	1	0	0
22121 23121	1	0	0

	Control	Profound - Severe	Moderate - Mild
4-7-2	n	n	n
22111 23211	1	0	0
4-7-4			
22222 23222	1	0	0
4-7-5			
22211 23411	0	0	1
4-9-1			
23111 21111	0	1	0
23121 21111	4	0	1
23122 21141	0	1	0
4-10-1			
23111 22121	1	0	0
23121 22111	0	1	0
23121 22121	1	0	0
23122 22122	1	0	0
4-10-3			
23222 22121	0	1	0
4-10-4			
23222 22221	0	0	1
4-11-1			
23111 23111	4	2	1
23111 23121	2	0	0
23121 23111	1	0	0
4-11-2			
23121 23211	0	0	1
4-11-4			
23211 23211	1	0	0

	Control	Profound - Severe	Moderate - Mild
4-11-4	n	n	n
23221 23221	0	0	1
4-11-5			
23111 23411	2	0	0
23411 23311	1	0	0
4-12-1			
23111 24121	1	0	0
23121 24121	0	0	1
4-12-5			
23111 24411	1	0	0
4-13-1			
24111 11111	2	1	0
4-13-5			
24321 21111	1	0	0
4-15-1			
24121 23111	2	0	0
4-15-5			
24111 23311	1	0	0
24111 23411	1	0	0
4-16-1			
24111 24141	0	2	0
4-16-5			
24411 24411	1	0	0
24411 24441	1	0	0
24421 24442	1	0	0

OTHER - GROUP 5

(3)

	Control	Profound - Severe	Moderate - Mild
13111 32111	0	0	1
14114 34414	1	0	0

(4)

	Control	Profound - Severe	Moderate - Mild
11111 41111	0	1	0
11111 41411	0	1	0
11411 41441	0	1	0
11122 41122	0	0	2
11411 44414	0	0	1
14111 41111	1	0	0
11111 44311	1	0	0
11411 44111	1	0	0
11131 43311	1	0	0
12122 41111	0	0	1
12111 41111	0	0	1
13211 42211	0	1	0
13111 42111	1	0	0
13121 43111	0	1	0
13111 44411	1	0	0
14121 41122	0	1	0
14431 41411	1	0	0
14111 43311	1	0	0
14411 43411	1	0	0
14111 44111	1	1	0
14111 44114	0	1	0
14111 44411	1	0	0
14411 44111	1	0	0
14421 44411	0	1	0
14431 44411	1	0	0
14444 44444	1	0	0

(7)

	Control	Profound - Severe	Moderate - Mild
24121 34121	0	0	1
24441 34441	0	1	0

(8)

	Control	Profound - Severe	Moderate - Mild
21111 41111	0	0	1
24124 44121	1	0	0
24444 44444	1	0	0

(10)

	Control	Profound - Severe	Moderate - Mild
34121 24421	0	1	0
34411 24444	1	0	0

(13)

	Control	Profound - Severe	Moderate - Mild
41411 14444	1	0	0
42111 13121	1	0	0
43411 11414	0	0	1
44111 11111	1	0	0
44114 11111	0	1	0
44444 13444	1	0	0
44141 14441	0	1	0
44321 14411	0	0	1
44444 14441	1	0	0
44411 14444	0	0	1

(16)

	Control	Profound - Severe	Moderate - Mild
41111 41111	1	0	0
41411 44111	1	0	0
42411 43311	1	0	0
43111 41111	1	0	0
43214 43214	0	1	0
44111 44111	4	0	0
44111 44411	1	0	0
44411 44411	0	1	0
44111 44141	0	0	1
44414 44144	0	0	1
44411 44114	1	0	0
44411 44422	0	1	0
44111 44431	1	0	0
44441 44442	1	0	0
44444 44444	0	1	1
44441 44444	0	1	0

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