Chapter 5

Using Dermatoglyphics from Down Syndrome and Class Populations to Study the Genetics of a Complex Trait

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Reprinted From: Fogel, T. 2004. Using dermatoglyphics from Down Syndrome and class populations to study the genetics of a complex trait. Pages 99-124, *in* Tested studies for laboratory teaching, Volume 25 (M. A. O'Donnell, Editor). Proceedings of the 25th Workshop/Conference of the Association for Biology Laboratory Education (ABLE), 414 pages.

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Introduction

The study of dermal ridges in the classroom is an effective way to introduce basic principles of quantitative genetics (see Mendenhall *et al*., 1989) and to demonstrate how the pattern of development caused by trisomy 21, can have multiple effects on the phenotype, including the pattern of dermatoglyphics.

This lab first appeared in the 11th volume of the ABLE proceedings. The present version has been revised and edited from the original and includes copies of the same Down syndrome prints. A section with student problems has been added to reinforce concepts. When I was asked to reprise the lab for the 25th anniversary of ABLE, I was, at first, skeptical. The science of dermal ridges is "old time" genetics compared to today's high-tech molecular biology. A literature search revealed surprisingly little genetic advance since the first publication of this lab in 1990. No dermatoglyphic genes have been mapped or cloned, for example. But the lab, despite its age, has many virtues. It still grabs students' attention. After all, what is more interesting than studying oneself! And it is an easy and fun avenue into the basics of quantitative genetics. As my colleagues in ABLE have pointed out over the years, the lab can be readily adjusted to different levels of biology or even simplified to a quick demonstration of human population variation.

Notes for the Instructor

The science of Dermatoglyphics, the study of ridged fingers, palms, toes, and soles, began with Sir Francis Galton in the late 1800s. His classification scheme for fingerprints was later adopted by Sir William Herschel in the late 1890s, as a new tool for individually identifying workers in India. By the beginning of the 20th century, dermatoglyphics had been adopted for forensics by Scotland Yard. Anthropologists seized on dermatoglyphics as another character they could quantify in human populations. And by the 1960's there was considerable research interest the relationship between genetic abnormalities and dermatoglyphic patterns (see Penrose, 1969; Reed, 1981).

Dermal ridges originate in the 6-7 week of development from volar pads composed of mesenchymal tissue. Ridges become visible by the 12th week. By the 24th week, development of the dermal ridge pattern is largely complete. The size, position, and shape of the volar pad are largely responsible for the ridge patterns observed. In general, small pads produce arches and larger pads produce whorls or arches.

The "Ridges and Furrows" website <*http://www.ridgesandfurrows.homestead.com/index.html*> has a great section on how dermal ridges form.

It has been observed that ridges are influenced by blood vessel nerve pairs at the border between the dermis and epidermis during prenatal development. Features such as inadequate oxygen supply, abnormal nerve growth, unusual patterning or distribution of the sweat glands, alternation of epithelial growth, or other features could influence ridge patterns. Because growth is a dynamic process, one in which many components contribute and mutually interact, there must be many genes involved.

Holt (1968) demonstrated a close correspondence between the observed and predicted correlation for total ridge count (TRC) and the degree of genetic relatedness. That is, TRC acts as an additive genetic trait with little dominance deviation. TRC fits the classical polygenic model in which one assumes that each gene "adds" in some small way to the total observed variability. But what is being "added?" Clearly, a phenotypic expression which requires a multitude of tissue types, all simultaneously undergoing developmental change, is at odds with a simplistic image of genes being somehow "additive." However, the additive gene model is not physically real to any organism. It is a mathematical tool that relates genotype to phenotype in a global sense. Put another way, understanding additive genetic variability yields predictive value about phenotypes in future generations, but does not tell you anything about the underlying biology. Why is this distinction important? Because it is critical that undergraduates understand the context and complexity of the genotype-phenotype relationship beyond just focusing on the statistical aspect of quantitative inheritance.

Dermatoglyphic analysis lends itself to a variety of thought questions that can help students link quantitative genetics and development.

1. Since dermal ridges are fixed for life, does that mean there is no environmental effect?

   Depends on what you mean by environment. Although the basic pattern and ridge count does not change as you age, scars can distort a pattern and heavy manual labor can wear them down. There is a developmental effect on the depth of ridges. Babies and the elderly have shallow ridges.

2. If all 10 fingers derive by mitosis and share a common set of genetic information (totipotency), why don't they all look the same?

   Prenatally, there appears to be several components influencing growth fields of ridge counts. Roberts (1979) partitioned variance components to argue for at least four major induction factors that seem to mirror observed developmental fields (1) some feature which influences the overall size and therefore ridge count, (2) another that increases ridge counts more on the radial than ulnar side of the finger, (3) a factor influencing the counts of the medial digits, (4) a factor influencing the lateral digits. Roberts concludes that, "each finger [acts] as a different point in relation to others in the multidimensional space produced by these factors." Put another way,

each finger is a different microenvironment of development.

3. If the left and right hands are both the products of a common genotype, why is there often a different ridge pattern for the same finger of each hand?

   Although there is a genetic component to ridge patterns, it is not purely deterministic. There must be stochastic effects during development that influence ridge patterns.

4. If we were to look at the dermatoglyphic patterns and ridge counts of identical twins would they be absolutely identical?

   Identical twins share extremely similar dermatoglyphic patterns and total ridge counts. Newman (1930) reported a +0.95 correlation for the total ridge count of identical twins compared to +0.46 for dizygotic twins. The similarity between identical twins suggests that there is a genetic effect on dermatoglyphic patterns. The small differences indicate stochastic effects.

5. What effect does the maternal environment have?

   Correlations between mother/child and father/child for TRC are similar, suggesting the maternal environment is not influential.

6. Why are dermatoglyphic patterns altered with Down Syndrome?

   The presence of the extra chromosome dramatically alters prenatal development and can have verying effects on each organ system. Generally, the nervous system is the most sensitive of the organ systems during prenatal development and characteristically leads to mental retardation for those with Down Syndrome. Not surprisingly, a trait such as ridge patterning which is influenced by field inductions, positioning of the volar pads, and nerve-vessel pairing would be affected in some complex, perhaps mutually interacting, fashion.

7. If Down Syndrome is a genetic trait caused by an extra 21, why is there individual variation in the ridge pattern and ATD angle?

   For each individual with Down Syndrome, trisomy 21 interacts with a unique genetic background that also influences expression.

8. Given that Down Syndrome affects many organ systems during prenatal development, should not other genetic defects alter dermal ridges for the same reason?

   Yes. Altered patterns are known, for example, for trisomy 18, trisomy 13, XXX, XXY, as well as for many duplications and deletions. A search on Online Mendelian Inheritance of Man *<http://www3.ncbi.nlm.nih.gov/Omim/searchomim.html>* using dermatoglyphics as a search term reveals many single gene traits that have distinctive dermatoglyphic features.

9. If nerve-vessel pairing influences ridge patterning, should not abnormalities of these tissues affect dermal ridges?

   Ridge aplasia is a rare autosomal dominant condition. No dermal ridges are formed on the hands or feet, a condition caused by a failure of nerves to grow into the epithelium during prenatal

development. Many serious congenital defects result in abnormal dermatoglyphic patterns termed ridge dysplasia (perhaps caused by deviations in the normal verve branching) or ridge distortion (likely the result of a disturbance in the spatial arrangements of nerves). Such clinical cases serve to demonstrate the multiplicity of effects (pleiotropism) a genetic condition can produce.

Materials

There are several methods for obtaining prints. Both work well. This exercise is written for use with the inking method. Porolon inkpads are available from Sirchie Finger Print Laboratories, Inc. (*www.sirchie.com*), catalog # FPT265. Do not use office-type inkpads. The ink is thin, hard to remove from your hands, and does not yield quality prints.

An alternative method for preparing fingerprints is to use three-quarter inch clear plastic tape. Rub graphite powder (available from chemical supply companies and hardware stores) over the area to be printed and press the tape gently against the surface. Peel off the image of the print and transfer onto paper. As a substitute for graphite powder, rub a pencil onto paper to deposit a thin layer of graphite. Rub fingers in the graphite and use tape to transfer the print to paper. To transfer palm prints or whole handprints the four-inch tape known as Book-Lock used by librarians to support the binding of paperbacks works well because it is thick and easy to handle. Other types of wide tape, such as packing tape, are too sticky and curl easily. Book-Lock can be obtained in 20-yard rolls *(www.thelibrarystore.com)*. One roll is good for about 80 handprints.

Other supplies needed are protractors (to measure atd angles) and dissecting scopes or hands lenses (for making ridge counts). Master copies of the Down Syndrome prints are provided in the Appendix. Cover Down's prints with clear plastic (or laminate) and use a felt tip pen to mark them up without disturbing the print.

Student Outline

The study of ridged skin – dermatoglyphics – was pioneered by Sir Francis Galton in the late 19th century. Since that time, extensive work has been carried out on the biology and genetics of skin patterns.

You will analyze fingerprint patterns, the total ridge count, and atd angles among members of the class and investigate dermatoglyphic differences found on individuals having Down syndrome (trisomy 21).

Fingerprint Patterns

Dermatoglyphic patterns are due to the convoluted layers of cells of the epidermis. At the peak of the ridges are found the pores of the sweat glands (see Figure 1). Ridges are laid during embryonic growth and patterns remain unchanged during postnatal development. However, extensive physical labor can wear down ridges and scars can distort the pattern. Also, ridge depth is influenced by age; both young children and older adults have thinner ridges.

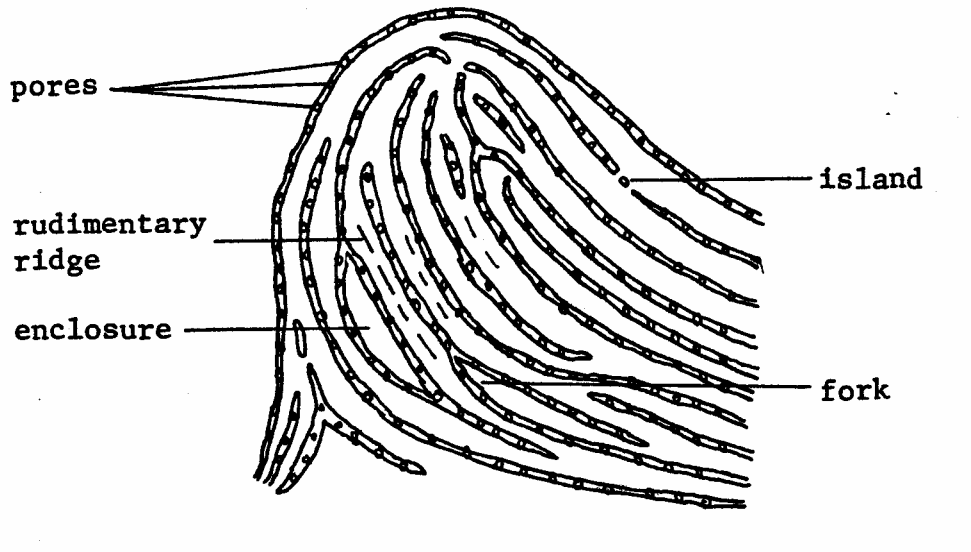

Figure 1. The detailed structure of dermal ridges.

There are three major classes of fingerprints - arches, loops and whorls (see Figure 2). Loops can be defined as radial or ulnar. Ulnar loops open towards the little finger and radial loops open towards the thumb. A useful descriptive term in dermatoglyphics is the triradius. A triradius is a point of convergence for three regions that separate almost parallel ridges. Loops have one triradius (on the thumb side if ulnar and towards the little finger if radial) and whorls have two. Arches lack a triradius. The population frequency for patterns is: radial loops 5.4%, arches 5%, whorls 26.1%, and ulnar loops 63.5%.

The genetics of fingerprint patterns is not well established. Based on one large pedigree, single gene control was deduced for certain patterns (Slatis et al., 1976). The lack of confirmation from other investigators and the cautious presentation of these results by the authors suggest that this scheme remains unconfirmed. Slatis and his colleagues suggest that all ulnar loops are the baseline pattern and that certain genes modify this phenotype. They also suggest that epistasis between loci may play a role.

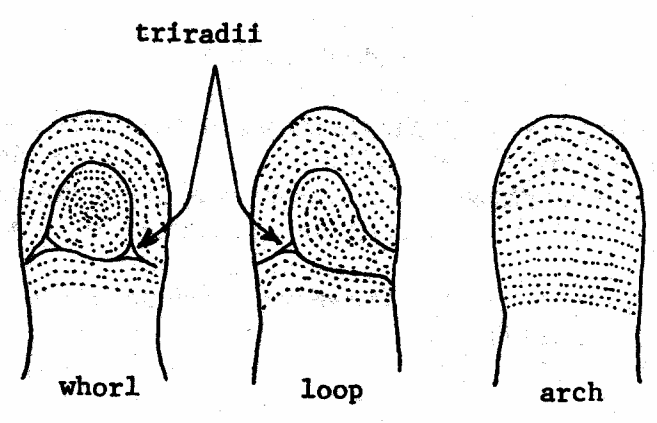

Figure 2. The three main types of fingerprint patterns. Whorls have two triradii, loops have one, and arches none.

Total Ridge Counts

A ridge count is made by drawing a line from the triradius to the center of the pattern and determining the number of intersected ridges between these two points (see Figure 3). Arches are defined as having a ridge count of zero. The ridge count of a whorl consists of the higher of the two counts. A total ridge count (TRC) is the summation of the ridge count for all 10 fingers. Other important considerations are quoted from Holt (1968):

> "The triradius is not included in the count, nor is the final ridge when it forms the centre of the pattern. Ridges which run close to the line without meeting it are excluded, but two ridges resulting from a bifurcation are both counted. It is usual to exclude from the count the fine secondary intervening ridges which occur occasionally, chiefly on thumbs. These secondary intervening ridges do not carry sweat gland pores. Furthermore, they are not as high as other ridges and whether or not they appear on a print depends on the degree of pressure exerted when the print is made. It is possible, therefore, to have two prints of the same finger, one showing secondary ridge and the other not. Islands, on the other hand, are always counted."

The genetics of total ridge counts has been studied in depth. It has been found that there is considerable variation among unrelated individuals, but there is a statistically significant positive correlation among relatives. This means that closely related individuals are more likely to be similar than distantly related ones due to the degree of shared genetic heritage. Detecting positive correlations among relatives is a common approach for analyzing the genetic component of complex phenotypic traits. With this method, it is very difficult, if not impossible, to determine the number of contributing genes. It is usually presumed that there are many genes controlling the overall phenotype (polygenes). Traits such as total ridge count that have a range of phenotypic expression are called quantitative traits. They are generally described in statistical terms (such as correlations) since the genotype is not known. However, in one study of total ridge counts, it has been suggested that half of the variation is controlled by a single locus (Spence et al., 1973). Possibly a number of other genes have some small effect on the phenotype also.

Figure 3. The technique of ridge counting. This loop has a ridge count of 13.

Palm Prints

At the base of the palm, there are usually four triradii called a, b, c, and d (see Figure 4). An axial triradius, called t, is usually located near the point where the palm is connected to the wrist. Two percent of normal individuals have this triradius positioned near the center of the palm (termed t"). A triradius found halfway in between these two positions (t') is found on twenty-one percent of the normal population. Approximately eleven percent of the population will have some combination of more than one axial triradius.

A useful descriptive measure is the atd angle formed by drawing a line from a to t and from d to t. If there are multiple axial triradii, only the largest angle is considered. The normal atd angle averages 39^o among females and 43^o among males. People with Down syndrome have an atd angle averaging 81^o. Elevated atd angles are also found on individuals with other forms of chromosomal abnormalities including trisomy 18, trisomy 13, Klinefelter syndrome (XXY) and Turner syndrome (XO). A wide range of unusual dermatoglyphic features other than the atd angle are also useful for recognizing genetic abnormalities. Dermatoglyphics is an especially quick and simple diagnostic tool for newborns. It is important to point out that many anomalies produce similar skin patterns so that corroboration with cytogenetic analysis or other appropriate tests is essential.

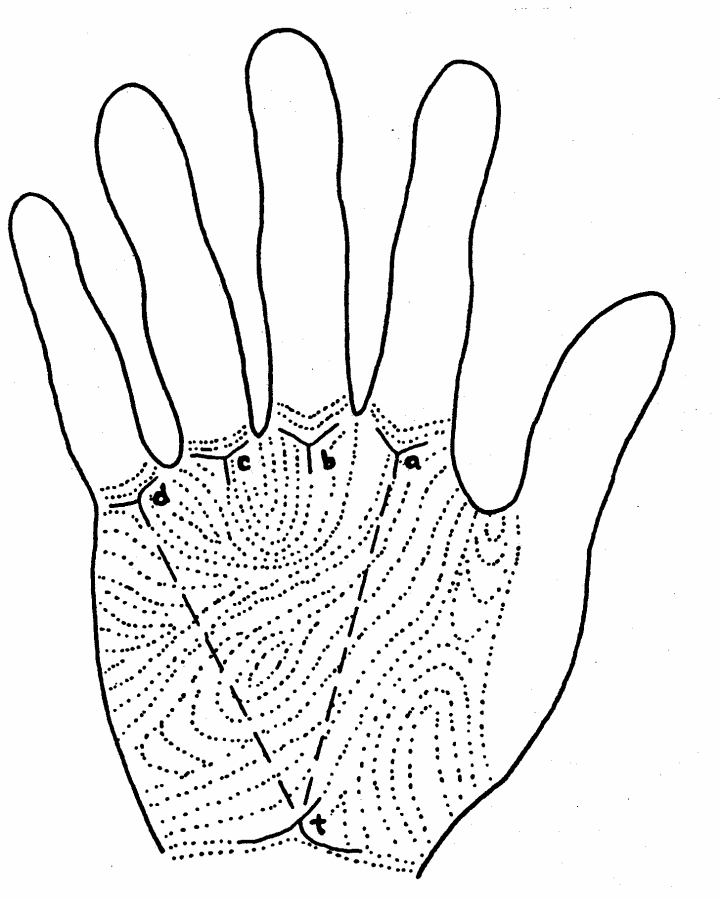

Figure 4. A palm print showing the triradii and ATD angle.

Dermatoglyphics of Down Syndrome

In addition to the elevated atd angle described above, several other features are also useful in the diagnosis of Down Syndrome. These include:

1. Fingers with mostly ulnar loops (especially the index finger); radial loops on the ring and index fingers; high incidence of ulnar loops (83%) compared to the normal population (63%).
2. A single flexion crease on the little finger. A flexion crease is the skin fold that occurs when you bend your fingers. Found in 26% of Down individuals; very rare in normal population.
3. Simian crease. Found in 53% of Down individuals; 2% in normal population.
4. A loop between the base of the index and middle finger and/or the middle and ring fingers. The loop opens to the inter- digital space.
5. Hypothenar ulnar loops, whorls or carpal loops. The palm is roughly divided into two halves: the thenar region on the thumb side and hypothenar on the little finger side. Carpal region refers to the very base of the palm.

No one feature is diagnostic and many of these patterns are found in the normal population. However, when several are present together, they are indicative of Down syndrome. On the print given to you look for the above-mentioned traits. Because the Down's prints vary in quality, it is not possible to determine if all of these traits are present on any one individual.

How to Make Fingerprints and Palm Prints

To make good fingerprints, touch the ink with your finger on its side and roll smoothly across the ink l80^o to the opposite side of the finger and lift. Repeat the motion on paper, transferring the print. It is easiest if you place the paper at the edge of the table to give your hand space to rotate. You will find that sometimes the second or third print after inking yields a clearer image of the ridges. Check the print to see if it is smudged. The triradii of loops and whorls must be visible. Repeat as necessary.

Palm prints require a similar procedure. The palm needs to be slowly rolled to transfer the edges. The a, t, and d triradii need to be visible. Remove ink with soap and water.

Students' *t* test

The *t* test is a statistical procedure for comparing two means ($\bar{x}_1, \bar{x}_2$) and will be used to detect a difference, if any, between the atd angles of the normal (classroom) population and that of the Down's population. Simply taking the difference between two means is insufficient. A mean can be composed of numbers that are clustered together or very dispersed.

To make a comparison, there must be a measure of dispersion around the mean. This is reflected in the variance - a value that is the average squared difference from the mean. With atd angles, the variance is a pooled estimate of both the normal (classroom) and Down syndrome population. It is presumed that the variance is the same for both. Keep in mind that the mean and variance are estimators; that is, they give an approximate value of the true population mean and variance from the data at hand.

The variance, s^2, *is:*

$$s^2 = \frac{\sum (x_{1i} - \bar{x}_1)^2 + \sum x_{2i} - \bar{x}_2)^2}{n_1 + n_2 - 2}$$

(n = sample size)

By rearrangement, the variance can be put in a form that is easier computationally:

$$s^2 = \frac{\sum x_{1i}{}^2 - \frac{\left(\sum x_{1i}\right)^2}{n_1} + \sum x_{2i}{}^2 - \frac{\left(\sum x_{2i}\right)^2}{n_2}}{n_1 + n_2 - 2}$$

The square root of the variance (termed the standard deviation) is used in the calculation of the *t*-value.

$$t = \frac{\bar{x}_1 - \bar{x}_2}{s\sqrt{\frac{1}{n_1} + \frac{1}{n_2}}}$$

Notice that the difference between the means (numerator) is adjusted by the degree of variation around the mean (denominator). If the denominator is small then division by a small number makes the *t*-value large. Statistical differences are detected when the calculated *t*-value surpasses the critical *t*-value on Table 1. The critical values establish a rejection level at $P<0.05$ for the appropriate degrees of freedom (degrees of freedom = $n_1 + n_2 - 2$).

Large sample sizes improve the estimators and the ability to detect a difference between two populations. Inspection of the formula for the *t*-value reveals that as n_1 and n_2 become larger, $1/n_1$ and $1/n_2$ become smaller. This has the effect of inflating the *t*-value and therefore enhances the ability to resolve differences between means.

Drawing conclusions with a *t* test requires a probability statement. There is no assurance that the conclusion is true; rather one argues that the statistical analysis supports a given outcome at a particular probability level.

Table 1. Critical values of t.

Degrees of Freedom	Value of t	Degrees of Freedom	Value of t
1	12.706	16	2.120
2	4.303	17	2.110
3	3.182	18	2.101
4	2.776	19	2.093
5	2.571	20	2.086
6	2.447	21	2.080
7	2.365	22	2.074
8	2.306	23	2.069
9	2.262	24	2.064
10	2.228	25	2.060
11	2.201	26	2.056
12	2.179	27	2.052
13	2.160	28	2.048
14	2.145	29	2.045
15	2.131	30	2.042

Procedure

1. Obtain a fingerprint pad and practice getting clear prints on a piece of scrap paper.
2. When you feel you have the knack, make a set of prints for each hand on separate sheets of paper. Be sure to label each sheet as to "left" or "right" hand.
3. Practice, then prepare palm prints on sheets of paper.
4. Draw a line from the triradii to the center of the pattern and use a magnifying lens or dissecting microscope to determine the ridge count. Sum the ridge count for ten fingers to produce a total ridge count.
5. Draw lines on your palm prints to form an atd angle and determine the angle with a protractor.
6. Obtain the class data for atd angles. Your instructor will provide laminated palm prints from Down syndrome individuals; use a felt tip pen to determine the atd angles and pool the class results.
7. Using the atd angles from the class and Down syndrome individuals perform a *t*-test and determine if claims for a difference exist.

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Problems in Quantitative Genetics

1. Complete a t-test on Down vs Class data for ATD angles. Report your interpretation of the results.

2. Suppose total ridge count is due to five genes that interact additively; each a_2, b_2 c_2 d_2 e_2 allele adds to the TRC while the others do not. TRC for the mother is 100 with genotype $a_1a_2\,b_1\,b_2\,c_1c_2\,d_1d_2\,e_1e_2$ and for the father is genotype $a_1\,a_1\,b_1\,b_1\,c_1\,c_1\,d_1\,d_1\,e_1\,e_1$. Assume that the total range for TRC in the human population is from 0-200.

   a. How many units must each contributing allele add to TRC?_____

   b. What is the phenotype of the father? _____

   c. For the above couple, what range of phenotypes is possible in offspring? _____

   d. Which two phenotypes (TRC values) are most likely? _____ and _____

   e. Two families are studied which have a large number of children. Parents of family "A" have TRC values of 80 as do all their children. Parents of family "B" also have TRC values of 80, but their children show a wide range of TRC values. Propose possible genotypes consistent with these findings.

      Genotypes of Family A: _____ and _____

      Genotypes of Family B: _____ and _____

      Note: In reality, it is likely that more complex genetic interactions exist, and that random developmental affects also influence the final outcome. Even identical twins can have slightly different TRC.

2. Below are actual student data. Use the data you collected from lab, together with the data below, to construct a frequency histogram for TRC that has 10 units per bar on the X axis. That is, group the data as 0-10, 11-20, 21-30, etc. then plot it.

123, 136, 62, 107, 116, 126, 150, 108, 173, 133, 69, 53, 194, 155, 144, 180, 128, 142, 95, 178, 128, 142, 139, 167, 132, 127, 142, 122, 164, 236, 177, 178, 52, 155, 59, 73, 81, 202, 158, 42, 160, 109, 90, 54, 153, 157, 136, 83, 104, 72, 181, 175, 40, 80, 193, 70, 154, 173, 138, 150, 212, 116, 194, 160, 178, 81, 56, 27, 58, 157, 135, 100, 180, 156, 31, 198, 150, 139, 91, 82, 127, 70, 109, 115, 147, 100, 144, 107, 165, 138, 176, 170, 96, 109, 123, 198, 95, 91, 7, 107, 121, 165, 185, 175, 159, 88, 141, 125, 160, 125, 99, 179, 123, 121, 149, 33, 212, 92, 68, 93, 119, 179, 128, 142, 118, 158, 73, 162, 145, 74, 155, 126, 193, 81, 94, 157, 141, 153, 60, 151, 168, 115, 171, 114, 106, 138, 122, 153, 173, 162, 181, 190, 192, 126, 174, 102, 115, 159, 80, 108, 131, 64, 76, 100, 79

Does the histogram suggest an approximate bell-shaped distribution?_____

3. The data below are TRC values from the families of students. Prepare two graphs. One should be a graph of mom on the y-axis and dad on the x. Keep in mind that parents are most likely to be unrelated and therefore, from a genetics point of view, they represent two people drawn randomly from the population. The second graph should show the child on the y-axis and the mid-parent on the x-axis. The mid-parent is the average of the values for mom and dad. Position a best fitting straight line through the points.

Accompany the graphs with an explanation as to how the graphs differ and why they differ.

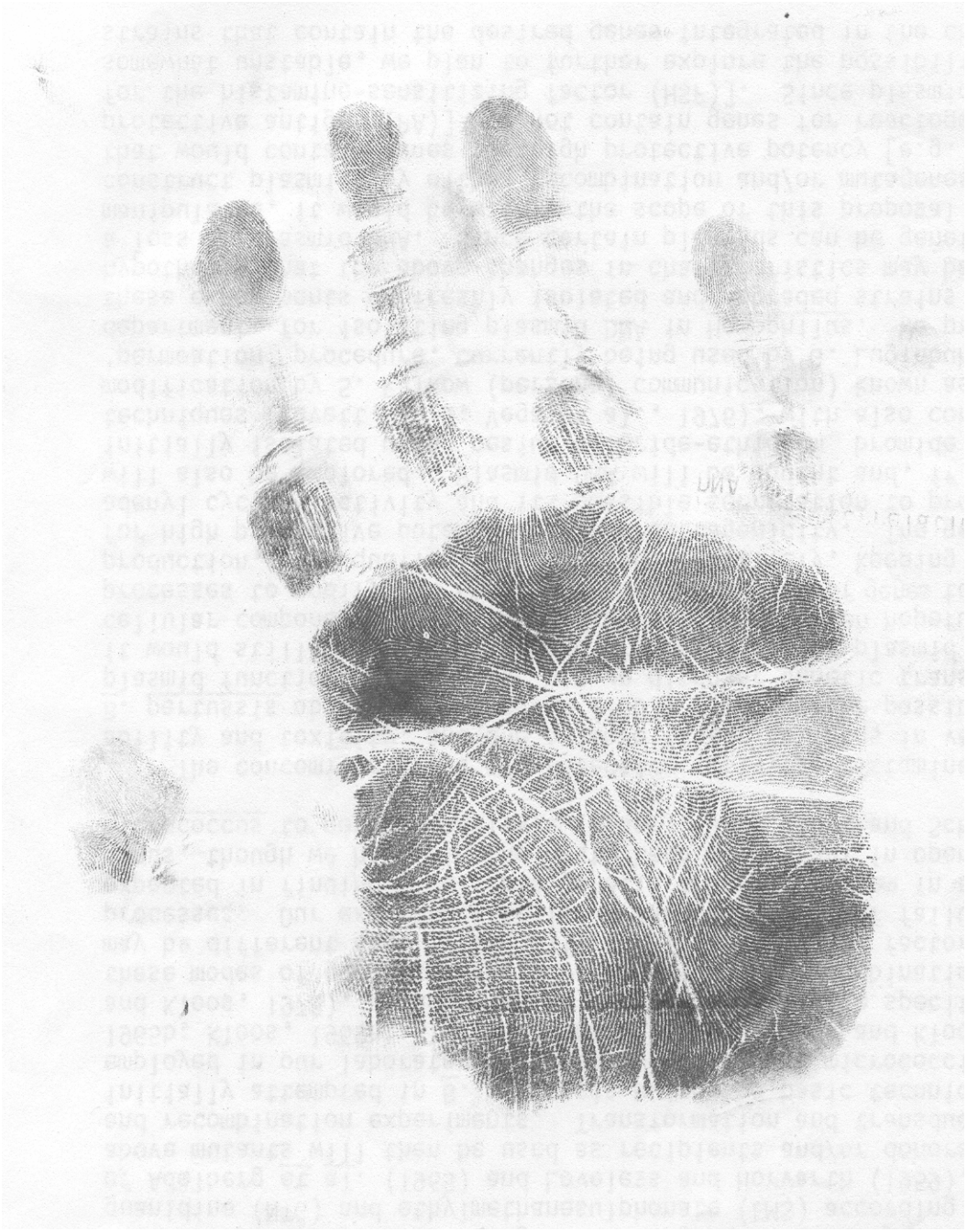
Mother	Father	Offspring
96	134	138
71	89	100
137	171	163
39	115	33
120	136	123
72	118	93
105	139	138
195	142	160
91	205	68
143	230	212
101	159	128
199	118	198
122	97	91
85	62	125
103	130	115
195	198	168
87	109	99
24	159	121
116	155	123

Appendix. Dermatoglyphic prints from six individuals with Down Syndrome.

Male, age 18, left hand

Male, age 18, right hand

Female, age 30, left hand



Female, age 30, right hand

Male, age 29, left hand

Male, age 29, right hand

Female, age 28, left hand

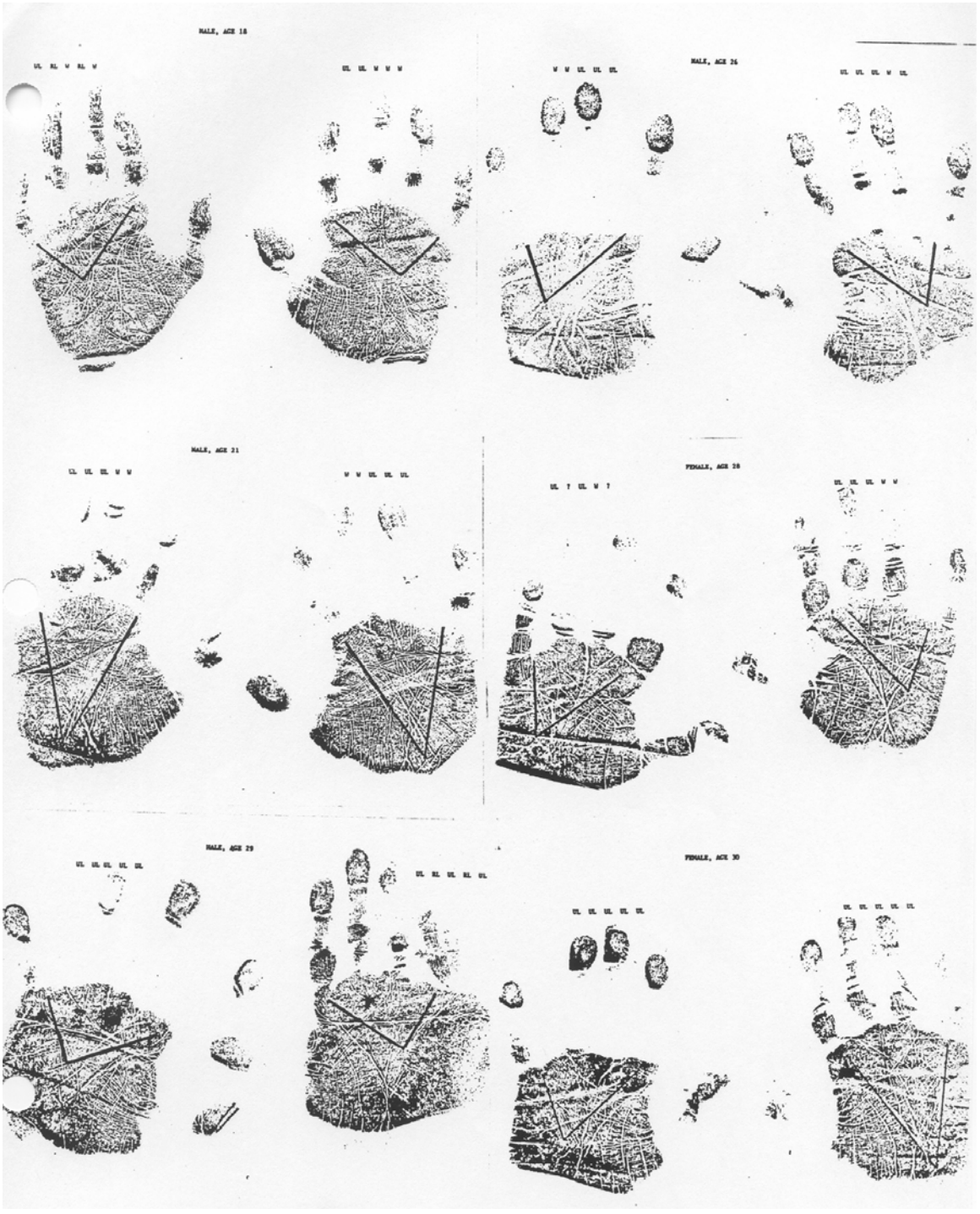
Female, age 28, right hand

Male, age 21, left hand

Male, age 21, right hand

Male, age 26, left hand

Male, age 26, right hand



Composite of handprints showing ATD angle and ridge patterns