Chapter 8

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

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Introduction

All too often when we teach genetics we limit ourselves to the "tried and true" single gene traits and "shy away" from more complex forms of genetic inheritance. After all, it is difficult enough to explain Mendelian Laws without the added complexity of quantitative variation. Ironically, understanding the relationship between genetic information and phenotypic expression at the organismal level has more to do with the interaction of many genes than it does to any one gene in isolation. Dermal ridges are an example of a trait that shows quantitative variation, is inherited, and is easily analyzed in the teaching laboratory. Further, sophisticated mathematical treatments need not be applied to teach the relationship between polygenic inheritance and expression.

The Student Outline is intended for students in an introductory genetics course. However, the exercise is easily adaptable to a beginning biology course and could even be shortened to less than one full laboratory period. The adaptability is possible by selectively eliminating one or more aspects of the laboratory experience. As written here, this exercise requires about 2–3 hours to complete. By deleting the statistical analysis of palm prints and using the Down Syndrome prints as demonstration, the exercise can be completed in less than 1 hour.

Materials

There are two methods for obtaining prints both of which will work adequately. This exercise is written for use with the inking method. Porolon ink pads can be obtained from: Sirchie Finger Print Laboratories, Umstead Industrial Park, P.O. Box 30576, Raleigh, NC 27612, (919) 781-3120. The cost, as of July 1989, is $19.50 US each (catalog no. FPT267). An alternative would be to purchase ink ($5.75/4 oz), a roller (FPT262A, 3 inches wide, $8.95), and supply your own surface (glass or steel); 2- and 4-inch rollers are also available.

I do not recommend using office-type ink pads. The ink is too thin and does not consistently yield high quality prints. Also, it is very difficult to remove from your hands once it has dried.

An alternative method is to use three-quarter inch clear plastic tape for fingerprints and 4-inch wide clear plastic tape for palm prints. Graphite powder (available from many chemical supply companies) is rubbed over the area to be printed and the tape is gently pressed against the surface. When the tape is peeled off an image of the print will be transferred to the tape which can then be pressed onto a sheet of paper. The 4-inch tape, known as Book-Lock, can be obtained in 20-yard rolls from: Library Store, P.O. Box 964, 112 E. South St., Tremont, IL 61568, (800) 548-7204. One roll will service about 40 students and costs $7.99 US (or less if purchased in bulk).

Other supplies for each student include a protractor (to measure the ATD angle) and a dissecting microscope (for making ridge counts). Also, the master copies of the Down Syndrome prints can be attached to clear plastic sheets so that students can mark them using felt-tip pens.

Notes for the Instructor

Dermatoglyphics and Development

Dermal ridges originate from fetal volar pads composed of mesenchymal tissue starting at the sixth to seventh week of development. The size and position of the volar pads are largely responsible for ridge patterns observed. In general, small pads produce arches and larger pads produce loops or whorls. Lateral displacement of the volar pad creates asymmetry of the pattern. Ridges become visible at about 3 months and are completed by the sixth month of prenatal development.

It has been postulated (see Schaumann and Alter, 1976) 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 sweat glands, alterations of epithelial growth, or other features could influence ridge patterns. Because growth is a dynamic process, one in which many components contribute and can mutually interact, there must be many genes involved.

Holt (1968) has demonstrated a very 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 is a near perfect fit to the classic 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 growing and changing as development proceeds, is at odds with a simplistic image of genes being somehow "additive." The resolution to this dilemma is to recognize that the additive model was never intended to be physically real to any organism. It is simply a functional mathematical tool that relates genotype to phenotype in a global sense. Put another way, understanding additive genetic variability gives you predictive ability about the phenotypes you might see 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 the complexity of the genotype-phenotype relationship beyond just focusing on the statistical aspects of quantitative inheritance.

For another example of teaching applications using dermatoglyphics see Mendenhall et al. (1989). A good overview of dermatoglyphics with clinical interpretations can be found in Reed (1981).

Thought Questions for Use in Class

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

Depends on what you mean by the 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.

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) argues for at least four major induction factors: (1) some feature which influences the overall ridge count, (2) another that increases ridge counts more on the radial than the ulnar side of the finger, (3) an increase in ridge counts more on the radial than ulnar side of the hand, and (4) an additional positive radial effect for the index finger and to a lesser extent on the thumb. Roberts (1979:488) 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. However, there are usually small, but detectable differences between them. This is a further confirmation that dermatoglyphic patterns and counts are under genetic control and also influenced by stochastic effects.

5. What effect does the maternal environment have?

Correlations between mother/child and father/child for TRC are similar suggesting that maternal the 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 varying 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 induction fields, 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 chromosome 21, why is there individual variation in the ridge pattern and ATD angle?

One would not expect an identical phenotypic outcome because each individual inherits a unique set of genetic information that also influences expression in addition to the overall effect produced by the extra chromosome.

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

Yes. Altered patterns are known, for example, for trisomy 18, trisomy 13, XXX, XXY, and an assortment of duplications and deletions to name just a few.

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

Ridge aplasia is a rare autosomal dominant condition. No dermal ridges are formed on the hands or feet, a condition probably 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 nerve branching) or ridge distortion (likely the result of a disturbance in the spatial arrangements of nerves). These clinical cases serve to demonstrate the multiplicity of effects (pleiotropism) a genetic condition can produce.

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 8.1). Ridges are laid during embryonic growth and remain unchanged during postnatal development. However, extensive physical labor can wear them down and scars can distort the pattern.

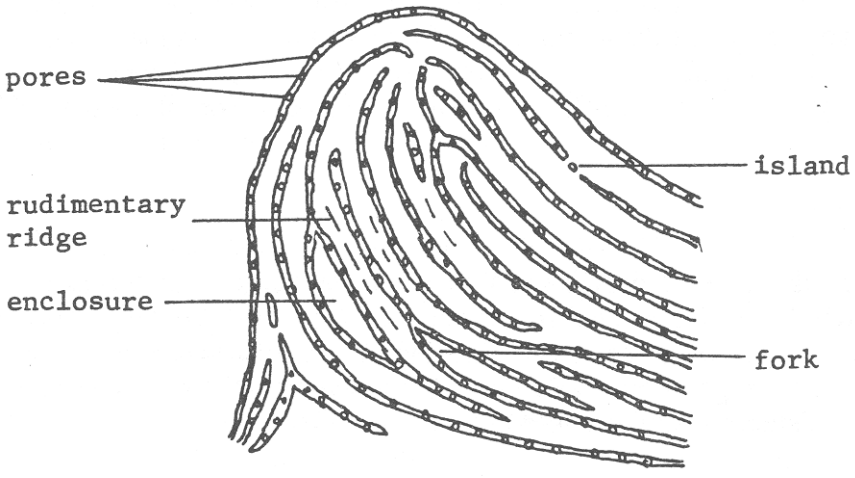
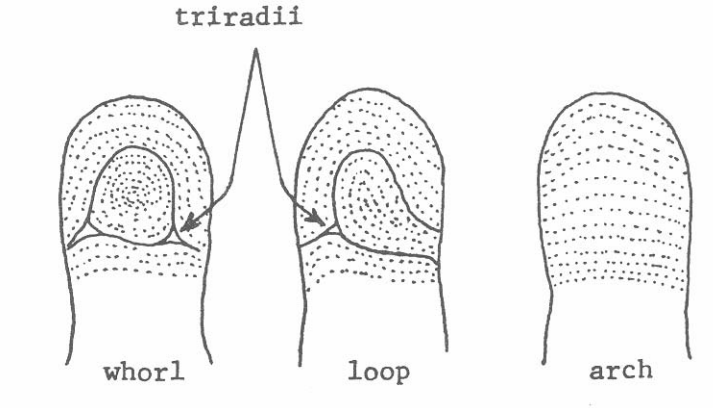


Figure 8.1 The detailed structure of dermal ridges.

There are three major classes of fingerprints: arches, loops and whorls (see Figure 8.2). 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.



F**igure 8.2.** The three main types of fingerprint patterns. Whorls have two triradii, loops have one, and arches none.

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 suggests that this scheme is tentative and awaits the test of time. 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. Table 8.1 is adapted from the work of Slatis et al. (1976).

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 8.3). 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 (1968:40):

"The triradius is not included in the count, nor is the final ridge when it forms the center 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 ridges and the other not. Islands, on the other hand, are always counted."

Table 8.1. The inheritance of fingerprint patterns.

Effect	*Inheritance*	*Genotypes*	*Phenotypes*
Thumb	Semidominant	*AA*	whorls, both thumbs
		Aa	2 ulnar loops or 1 ulnar loop and 1 whorl
		aa	ulnar loops, both thumbs
Thumb	Dominant	*B*-	arches on thumbs and often other fingers
Index finger	Dominant	*C*-	radial loops on index finger, often associated with an arch on the middle finger
Ring finger	Semidominant	*CC*	whorls, both fingers
		Cc	2 ulnar loops or 1 ulnar loop and 1 whorl ulnar loops on both fingers
Ring or little finger	Recessive	*dd*	radial loops
All fingers	Dominant	*E*	whorls on all fingers except for an ulnar loop on middle finger
All fingers	Dominant	*F–**	arches on fingers

* may be more than one gene involved

The genetics of total ridge counts has revealed 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 which 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 8.3 . 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: *a*, *b*, *c*, and *d* (see Figure 8.4). An axial triradius (*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 21% of the normal population. Approximately 11% 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 ATD angle averages 48° among normal individuals. People with Down Syndrome have an ATD angle averaging 81°. 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.

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.

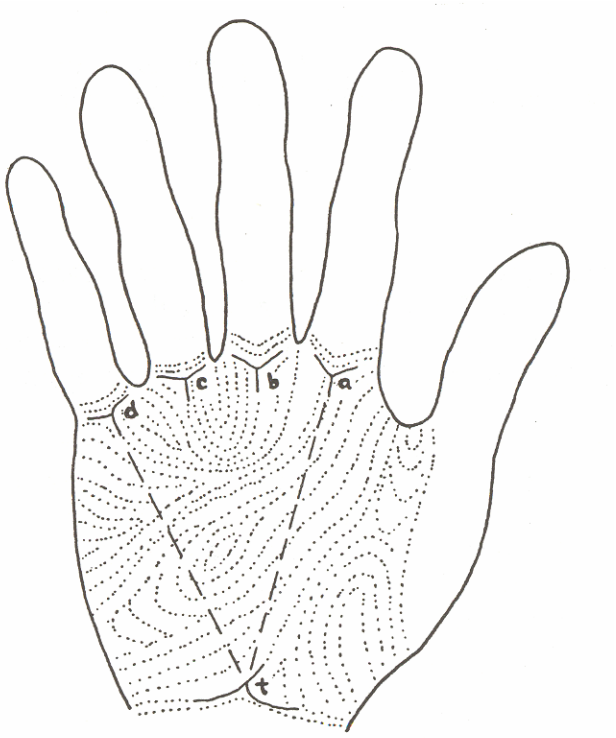


Figure 8.4. A palm print showing the triradii and ATD angle.

2. A single flexion crease on the little finger. A flexion crease is the skin fold that occurs when you bend your fingers.
3. Simian crease.
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 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° 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 from your hands with soap and water.

Students' *t* test

The *t* test is a statistical procedure for comparing two means and will be used to detect a difference, if any, between the ATD angles of the normal (classroom) population and that of the Down population. Simply taking the difference between two means does not convey enough information. 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 which is the average squared difference from the mean. In the case of 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:

$$s^2 = \frac{\Sigma (x_{1i} - _1)^2 + \Sigma (x_{2i} - _2)^2}{n_1 + n_2 - 2} \quad (n = \text{sample size})$$

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

$$s^2 = \frac{\Sigma x_{1i}^2 - (\Sigma x_{1i})^2/n_1 + \Sigma x_{2i}^2 - (\Sigma x_{2i})^2/n_2}{n_1 + n_2 - 2}$$

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

$$t = \frac{x_1 - x_2}{s\,(1/n_1 + 1/n_2)^{1/2}}$$

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 in Table 8.2. The critical values establish a rejection level at $P < 0.05$ for the appropriate degrees of freedom (degrees of freedom = $n_1 + n_2 - 2$).

If larger samples were available, the ability to detect a difference between two populations would be improved. 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 detect small 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 8.2. 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 got the knack, make a set of prints on the data sheets.

3. Practice, then prepare palm prints on the data sheet.

4. To determine your genotype, classify your fingerprints according to the scheme by Slatis et al. (1976).

5. Draw a line from the triradii to the center of the pattern and use a dissecting microscope to determine the total ridge count (TRC).

6. Obtain class data for total ridge counts and make a histogram with 10 ridge intervals. A quantitative trait often demonstrates a bell-shaped distribution if sufficient data points are available. Is there a trend towards such a distribution?

7. Draw lines on your palm prints to form an ATD angle and determine the angle.

8. Obtain the class data for ATD angles. Your instructor will provide palm prints from Down Syndrome individuals; determine the ATD angles and pool the class results. Do not write directly on these prints; lay clear plastic over them and use the felt-tip pens provided.

9. Using only the data for the right hand, calculate the class mean and the mean for Down Syndrome individuals. Use the left had print of Down Syndrome individuals if the right hand print is not measurable. Perform a *t* test and determine if claims for a difference exist. Have your analysis checked by your instructor.

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APPENDIX A

Data Sheet for Finger Print Patterns and Counts, and Palm ATD Angles

LEFT HAND

	Thumb	Second	Third	Fourth	Fifth
Pattern	_____	_____	_____	_____	_____
Ridge Count	_____	_____	_____	_____	_____

Data Sheet for Fingerprints

RIGHT HAND

	Thumb	Second	Third	Fourth	Fifth
Pattern	_____	_____	_____	_____	_____
Ridge Count	_____	_____	_____	_____	_____

Data Sheet for Palm Prints

	LEFT PALM	RIGHT PALM
atd angle	_____	_____

APPENDIX B

Statistical Analysis Sheet for Conducting t test Using Class and Down Syndrome Population Data

t-test Data Sheet for atd angles. This sheet is to be turned in along with a graphic distribution of total ridge count on the back side.

Data	Class (X_{1i})	X_{1i}^2	Down's (X_{2i})	X_{2i}^2
1				
2				
3				
4				
5				
6				
7				
8				
9				
10				
11				
12				
13				
14				
15				
16				
17				
18				
19				
20				

ΣX_{1i} = _____ ΣX_{1i}^2 = _____ ΣX_{2i} = _____ ΣX_{2i}^2 = _____

$\bar{X}_1 = \frac{\Sigma X_{1i}}{n_1}$ = _____ $\bar{X}_2 = \frac{\Sigma X_{2i}}{n_2}$ = _____

$\frac{(\Sigma X_{1i})^2}{n_1}$ = _____ $\frac{(\Sigma X_{2i})^2}{n_2}$ = _____

$$s^2 = \frac{\Sigma X_{1i}^2 - (\Sigma X_{1i})^2/n_1 + \Sigma X_{2i}^2 - (\Sigma X_{2i})^2/n_2}{n_1+n_2-2}$$

$$t = \frac{\bar{X}_1 - \bar{X}_2}{s\sqrt{1/n_1 + 1/n_2}} = _____ \quad df = n_1 + n_2 - 2 = _____$$

Conclusion _____

APPENDIX C
Left and Right Palm Prints from Six Down Syndrome Adults Showing the Position of the ATD Angle

The figure on the following page serves as the instructor key to go along with the six full-size prints provided on the succeeding pages. Above each hand is the fingerprint pattern established from prints that were taken at the same time (UL = ulnar loop; RL = radial loop; and W = whorl). Two fingerprints from the 28-year-old female were unreadable.

An adjunct to the Down Syndrome exercise would be to compare the frequency of pattern types between the class and the Down Syndrome individuals, the latter typically showing a higher frequency of ulnar loops.

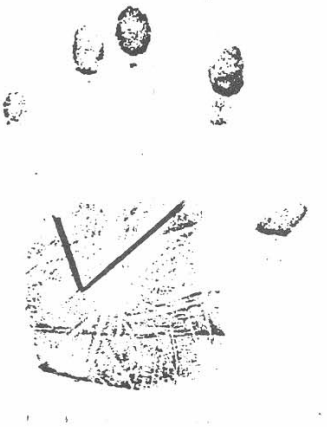
MALE, AGE 18

UL RL W RL W

UL UL W W W

MALE, AGE 26

W W UL UL UL



UL UL UL W UL

MALE, AGE 21

UL UL UL W W

W W UL UL UL

FEMALE, AGE 28

UL ? UL W ?

UL UL UL W W

MALE, AGE 29

UL UL UL UL UL

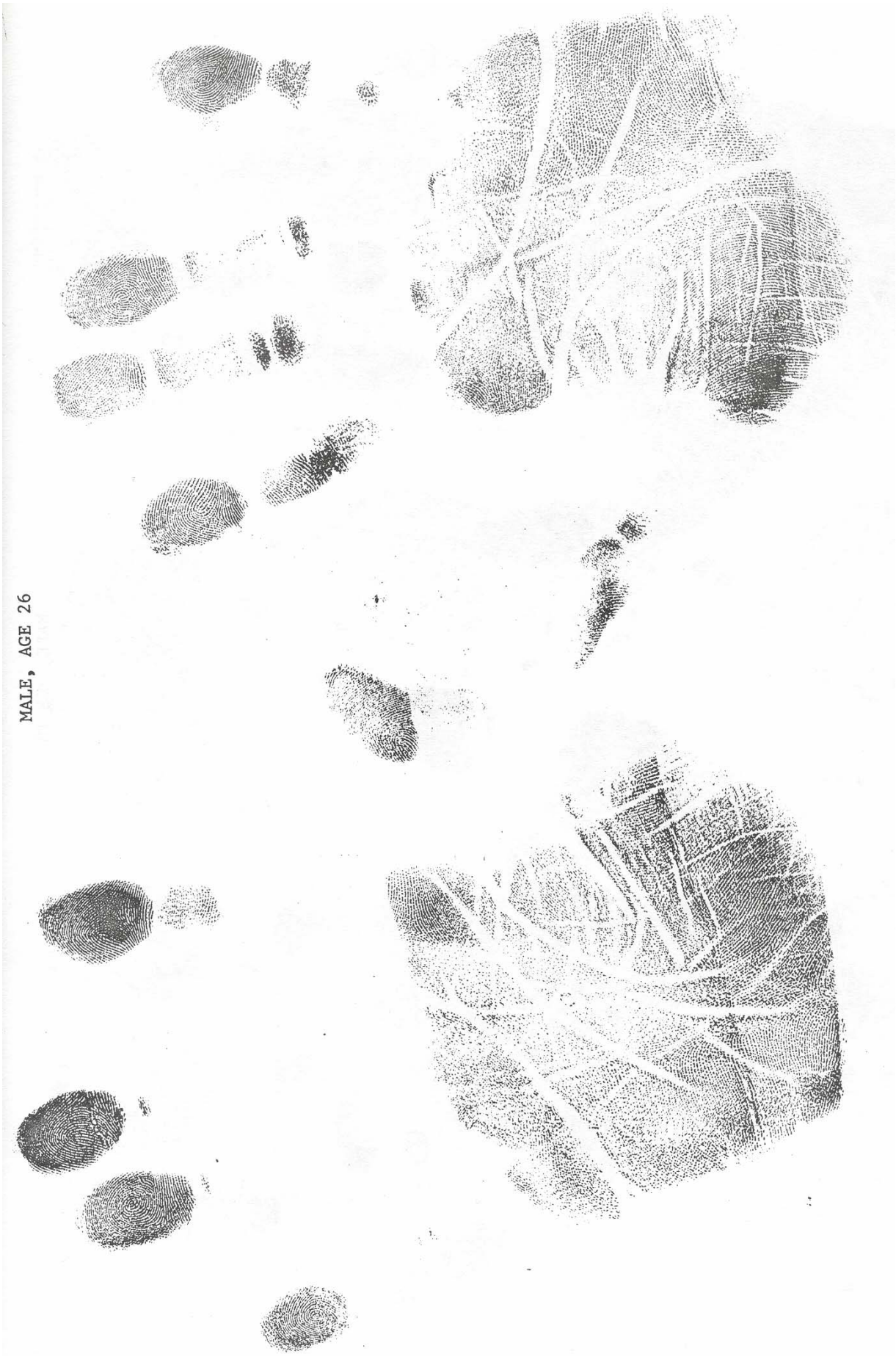
UL RL UL RL UL

FEMALE, AGE 30

UL UL UL UL UL

UL UL UL UL UL

MALE, AGE 18

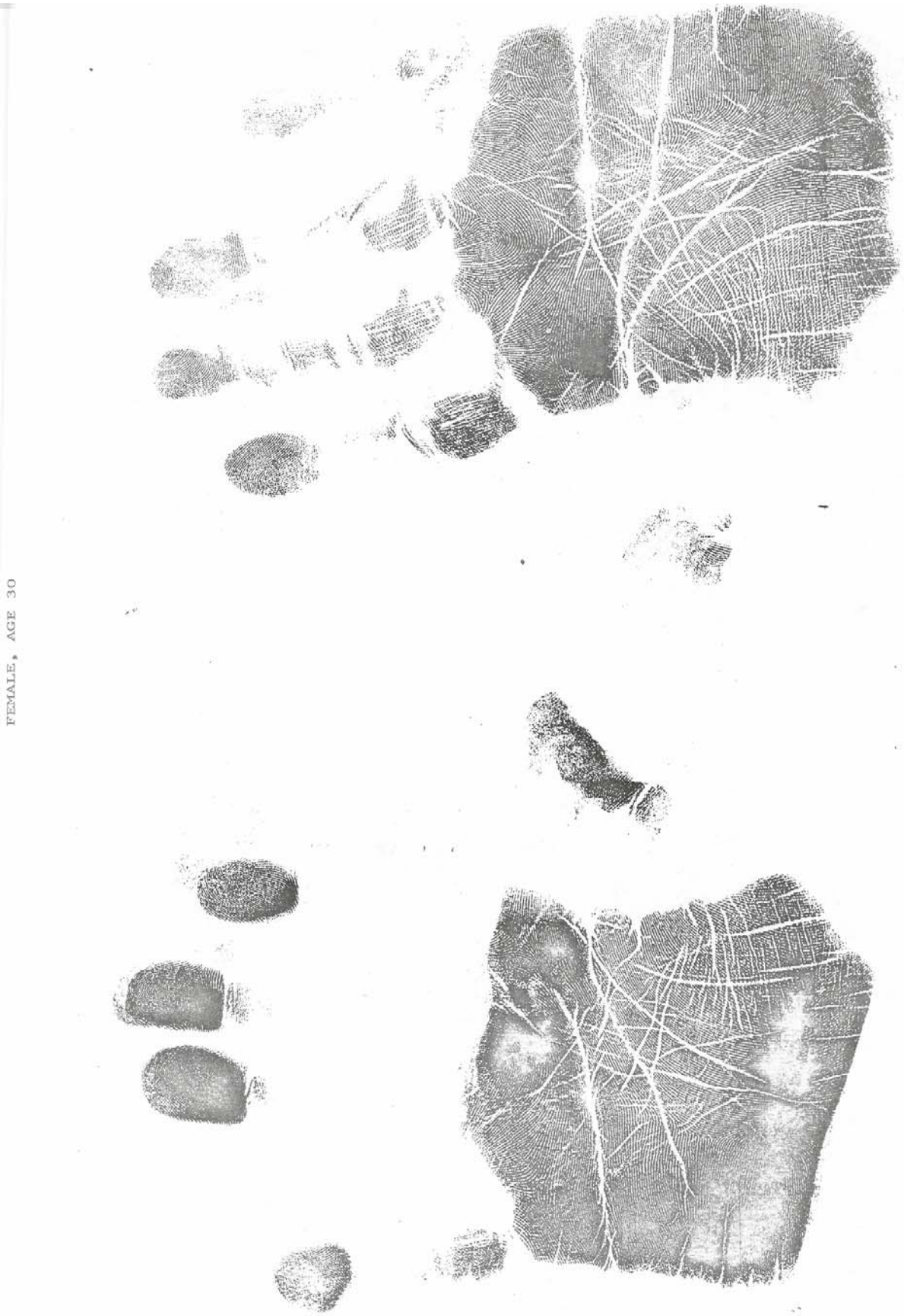


MALE, AGE 26

MALE, AGE 21

FEMALE, AGE 28

MALE, AGE 29



FEMALE, AGE 30