

DERMATOGLYPHICS AND KABUKI SYNDROME



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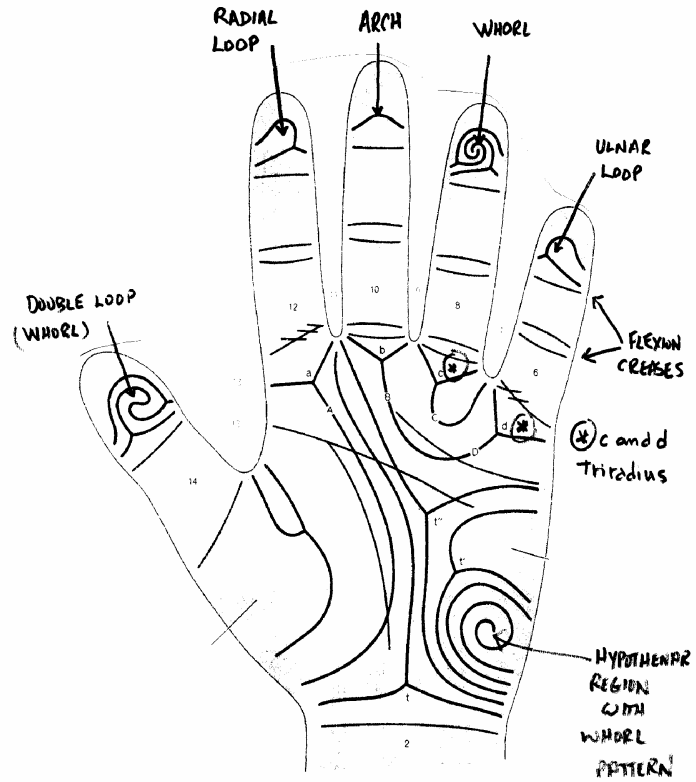
Dermatoglyphics (writing on the skin in Greek) is the study of epidermal ridges. Epidermal ridges form early in fetal life, and are unique to each individual. They consist of patterns of ridges on the finger pads, palms and soles of all individuals. They form different patterns, and are unique to individuals. This means they can be used for personal identification in criminal investigations. In genetics and medicine, they are useful in diagnosis, since recurring abnormal patterns are often seen in a variety of genetic syndromes. In addition, creases are formed on palms and soles that are also altered in syndromes. Although creases are not part of epidermal ridges, which require a magnifying glass or an ink impression to examine thoroughly, creases are part of what a geneticist looks at during his or her dermatoglyphic analysis and examination.

In Down syndrome, the creases are frequently abnormal on the palms with two of the three large creases forming what appears to be a single palmar crease (31% compared to 2% of controls). Also, individuals with Down syndrome have tibial arch patterns on the soles near the base of the great toes (60% compared to 0.5% of controls) and they tend to have 10 ulnar loops on their finger pads (30% compared to 7% of controls).

For Down syndrome, there is a diagnostic test, the chromosome analysis, that confirms the presence of 3 chromosome # 21's, instead of the usual 2. Therefore, dermatoglyphic analysis has become less important for the diagnosis of Down syndrome than for syndromes in which the genetic alteration has not been identified, such as for Kabuki syndrome (KS).

In many children with KS (over 75%), there are prominent fetal fingerpads. Usually these fingerpads become flat by the time of delivery, but in KS individuals, they remain prominent. This is not specific for KS, as they have been described in other syndromes, and can also be present in individuals without a genetic syndrome. Dr. Niikawa and co-authors brought our attention to the fact that in most people with KS, there are dermatoglyphic findings that separate affected individuals from unaffected. His findings showed that there was an increase in ulnar loops (63%); absence of digital triradius c (48%); absence of digital triradius d (30%); increase of hypothenar loops; and a single flexion crease of the 5th finger. Overall, in his study of dozens of KS individuals, about 93% showed some unusual dermatoglyphic findings. (See illustration).

Geneticists use dermatoglyphic analysis to help support the diagnosis of KS. However, as in Down syndrome, eventually the genetic cause of KS will be established, and the use of dermatoglyphic analysis will become less important.



This illustration shows the various landmarks related to dermatoglyphics and some common patterns or formations. In Kabuki syndrome, there are at least five commonly seen changes: (1) increase in ulnar loops (2) absent of the digital c or d triradius –region highlighted with asterix (3) increase in hypothenar patterns (4) single flexion crease in 5th digit (5) prominent fingerpads (not shown).