# A previously unreported syndrome of multiple scalp whorls and associated anomalies

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## Summary

A 13-month-old male infant with 14 hair whorls in the scalp, sparse frontal hair, wide forehead, ectropion, abnormal implantation of eyelashes, peculiar face and depigmented nipples is reported. Other aspects of his physical and mental development were within normal limits. The constellation of clinical features in this patient appear to represent a previously undescribed syndrome.

## Report

Hair whorls are a normal characteristic of human scalp hair although in black-skinned individuals with curly hair they may be difficult to identify. The vast majority of individuals have just one whorl; less than 5% have two whorls and three whorls is exceptional.

Here, a patient with 14 hair whorls all over the scalp and several other congenital malformations is described. The implications of this case for the developmental theories of hair whorl formation and the possible association between multiple hair whorls, brain development, and other anomalies are discussed.

The patient was a 13-month-old boy, the offspring of an uneventful pregnancy and uncomplicated delivery. At birth the patient had sparse hair with a wide forehead, and mild ectropion. Physical and mental development were normal. The family history was unremarkable: The father, mother, three brothers and one sister were all apparently normal.

Physical examination of the patient showed a peculiar face with wide protruding forehead, sparse eyebrows and eyelashes, bilateral ectropion, abnormal implantation of eyelashes and depigmented nipples. The scalp hair was thin, sparse in the frontal area, and had 14 whorls distributed all over the scalp. (Figs 1 and 2). Some whorls had a clockwise rotation and others, anticlockwise. Microscopic examination of hair was normal. Dermatoglyphics were not studied. Hair whorls in other body areas were not found.

A biopsy from the scalp showed normal skin with a reduced number of normal-appearing hair follicles and eccrine sweat glands. Routine blood tests, karyotype, and urine amino acids were within normal limits.

This case seems to be the first reported example of multiple hair whorls occurring all over the scalp. Over 95% of individuals have one posterio-parietal whorl located anterior to the posterior fontanelle. Most whorls are located on the right side of the scalp midline and have a clockwise rotation.[1–4]

Bernard *et al.*[5] in a study of 503 new-borns found that 75.4% of whorl patterns, whatever their type are located 2–4 cm from the posterior fontanelle. Scalp whorls are determined before the 18th week of gestation and the inclination of follicles is influenced by the pressure of the growing brain on the scalp. Craniocerebral anomalies may induce hair-pattern alterations.[3, 4]

'Mechanical tension' is the most widely accepted theory to explain the scalp hair slope and patterning: the pattern is thought to result from the tension placed on the epidermis during rapid expansion of the cranium while the follicle is growing downward into the loose underlying mesenchyme. The epicentre of the cranial whorl stretch would constitute the normal parietal whorl.[2]

However, cases with temporal or frontal whorls or with an occipital ridge[2] or cases such as ours, are difficult to explain by this mechanical tension theory. Another theory proposes a variable low or high metabolic activity in certain areas of the scalp that would

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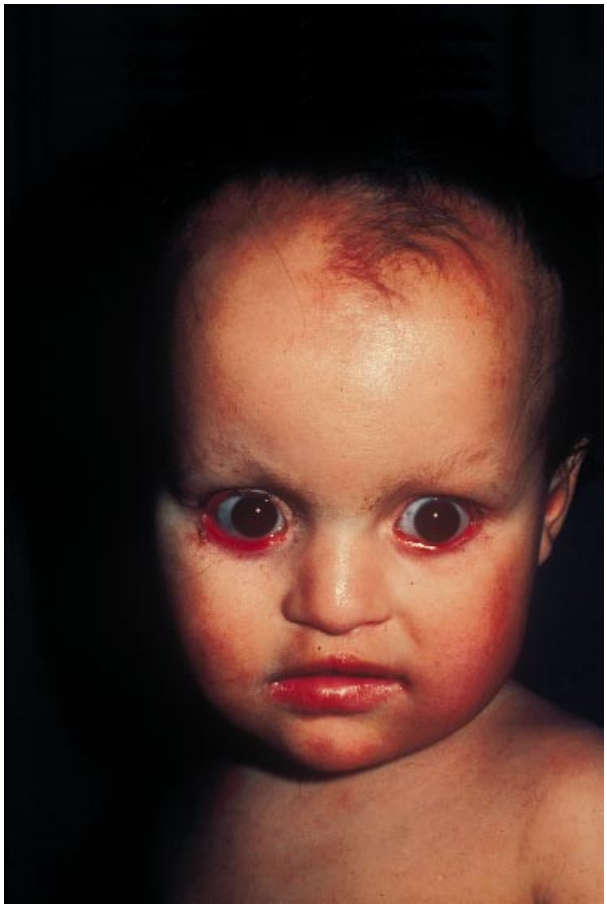

**Figure 1** Characteristic facies of the patient with sparse frontal hair, wide forehead, abnormal eyelash implantation and ectropion.

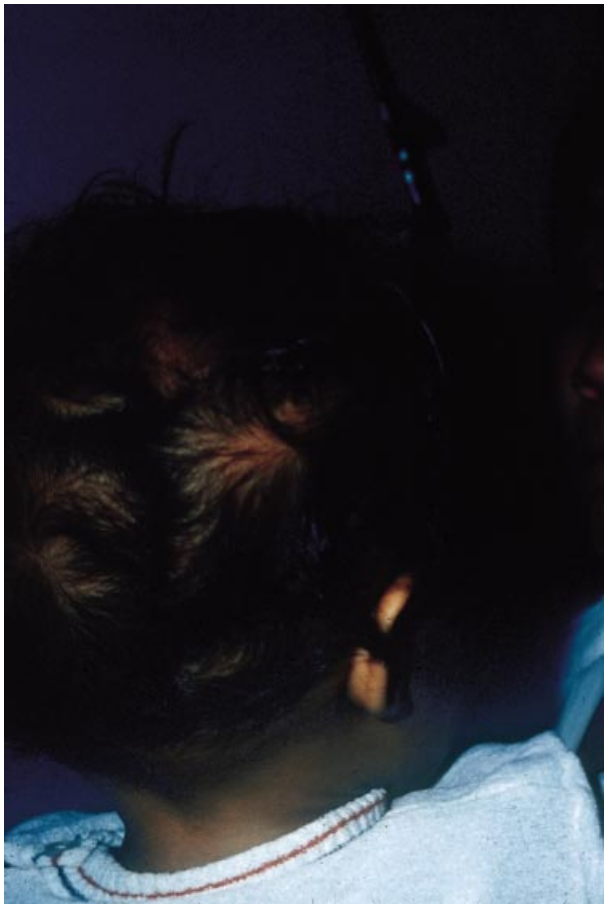

**Figure 2** Back of the head showing several hair whorls with clockwise and anti-clockwise rotation of hair.

result in a more active growth on one side of the follicle than on the other.[2] Direct evidence for this or a genetic basis for hair whorl patterns, however, have not being documented thus far.

The study of hair patterns, or trichoglyphics, has related aberrant scalp patterning to abnormal brain or skull development[1] and to a number of dysmorphic syndromes.[2, 3] Other authors, however, have failed to find a direct relationship.[4–6] The literature does not appear to contain any reports of multiple hair whorls (three or four) with associated anomalies. Moreover, a highly significant lack of multiple occipital whorls was found in mentally subnormal patients compared with normal controls.[3, 5]

Naevoid disseminated patches of hypertrichosis, in glabrous skin, associated or not to other anomalies should also be considered in the differential diagnosis.[7, 8]

In summary, this patient represents a most unusual example of disseminated multiple scalp whorls associating minor alterations and a peculiar face. Previously, an association of abnormal eyelash implantation (distichiasis) and ectropion has been reported as an autosomal dominat trait in three generations.[9] However, no hair abnormalities were documented in that pedigree, thereby establishing the constellation of clinical features in the present case as a unique entity. Reports of any similar cases may help to better characterize this rare condition.

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