GENETICS AND GENODERMATOSES

PHYLLOID HYPOMELANOTIC MOSAICISM: REPORT OF 6 NEW CASES

María Del Carmen Boente [(1)] - María José Fiandrino [(1)] - Norma Beatriz Primc [(2)] - Daniela Montanari [(3)]

Hospital Del Niño Jesús, Dermatology, San Miguel De Tucumán, Argentina [(1)] - Hospital Del Niño Jesús, Dermatology, San Miguel De Tucumán, Argentina [(2)] - Hospital Del Niño Jesús, Genetics, San Miguel De Tucumán, Argentina [(3)]

Background: Phylloid mosaicism is a distinct pigmentary infrequent type of mosaicism characterized by congenital hypo or hyperchromic symmetric macules resembling floral ornament. It is frequently associated with chromosomal abnormalities, mental retardation, agenesis of corpus callosum, conductive deafness, retinal colobomas, cranio-facial and musculoskeletal anomalies.

Observation: We present the findings of a research study of 6 female patients with phylloid hypopigmented mosaicism with ten years of follow-up study. Complementary studies (spinography, audiometry and ophthalmological controls) were carried out to analyze probable associations.
Two sisters in one of the families had hypopigmented leaf-like macules affecting hemilaterally the upper part of their torsos and necks. Two sisters from a second family had hypopigmented rounded and oval macules on their chests and necks. One patient had oblong, lanceolated and rounded lesions on her left upper limb, and the other patient had similar lesions on her right upper limb.
In none of the cases were chromosomal abnormalities detected.
The presence of scoliosis was found in the follow-up studies of five of the six patients. No other extracutaneous anomaly was found.

Key message: In our experience, phylloid hypomelanosis was more frequent in females. No chromosomal abnormalities were detected. The only systemic association was scoliosis.
It should be remarked that among these 6 cases there were 2 families with two affected sisters in each of them.