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Case Report

Hypomelanosis of Ito: a cutaneous disease with multisystem involvement

Jacinta Fonseca[1*], Karina Oliveira[1] and Ana Luísa Leite[1]

[1]Department of Pediatrics, Centro Hospitalar de Vila Nova de Gaia/Espinho, Vila Nova de Gaia, Portugal.

Summary

Disorders of hypopigmentation are frequent in a paediatric consultation. Selected diseases have signature skin findings which can identify a multysistemic disorder.

The authors report a case of hypomelanosis of Ito, a rare neurocutaneous syndrome, characterized by streaks and swirls of hypopigmentation following the lines of Blaschko associated to systemic abnormalities involving the central nervous system and musculoskeletal system.

Introduction

Hypomelanosis of Ito (HI) is a neurocutaneous phenotype characterized by hypopigmented anomalies along the Blaschko lines, with systemic abnormalities involving the central nervous and musculoskeletal systems. It was firstly described as incontinentiapigmentiachromians in 1952 [1]. Prevalence is unknown but incidence has been estimated at between 1/3000 and 1/10.000 [2]. The exact cause of HI is unknown. Many cases are associated with genetic mosaicism and sporadic gene mutations [1-4].

Case Report

We present a case of 12-year-old girl with deficits in attention, motor control and perception, mild mental impairment, thoracolumbar scoliosis, hemihypertrophy and dysmetry of the left lower extremity. For obesity and skin lesions was referred to pediatric endocrinology consultation. Physical examination revealed hypopigmented macular lesions in streaks and whorls distributed over the trunk, upper and lower limbs of the left side of the body, corresponding to the lines of Blaschko, from birth (Fig.1 A and B). The face, palm and soles of feet were

spared.Electroencephalogram (EEG) showed slower theta activity. Magnetic resonance imaging (MRI) of the brain revealed dilatations of the Virchow-Robin spaces (Fig. 2 A and B). Cutaneous findings, along with musculoskeletal and neurological symptoms were suggestive of the diagnosis of HI.

Discussion

Hypomelanosis of Ito, initially referred to as incontinentiapigmentiachromians, is a rare neurocutaneous disorder [1]. The skin phenotype is characterized by unusual unilateral or bilateral cutaneous macular hypopigmented whorls, streaks and patches, corresponding to the lines of Blaschko, that usually develop within the first two years of life.The lines of Blaschko are relatively consistent and distinct from dermatomal lines. They represent lines of orderly migration of mesodermal and ectodermal precursors during embryogenesis. Multiple organ systems can be involved including brain, musculoskeletal, cardiovascular, eyes, kidneys, and teeth [3-5].

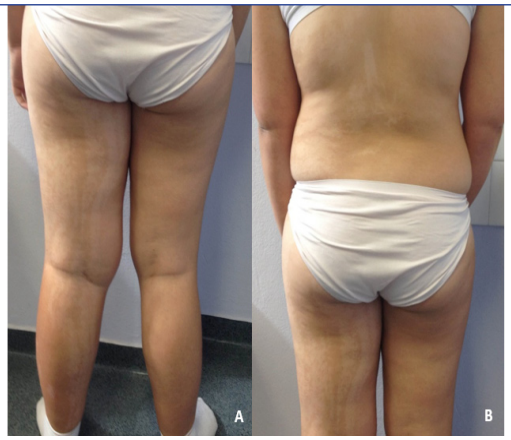

Figure 1: A. Hemihypertrophy of the left lower extremity with hypopigmented macular lesions in streaks and whorls. **B.** Hypopigmented macular lesions in streaks and patches over the left side of the body.

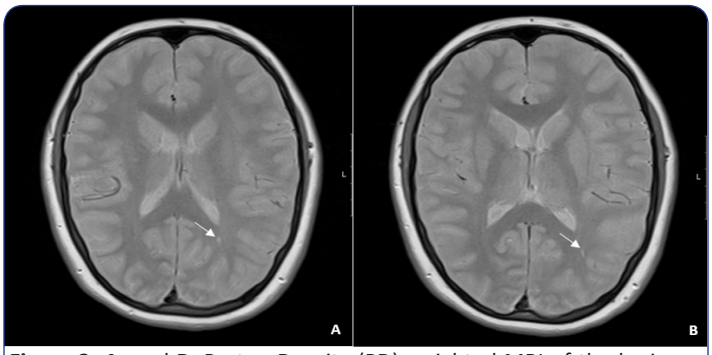

Figure 2: A. and **B.** Proton Density (PD) weighted MRI of the brain revealed dilatations of the Virchow-Robin spaces. EEG showed slower theta activity.

***Corresponding author:** Jacinta Fonseca, Department of Pediatrics, Centro Hospitalar de Vila Nova de Gaia/Espinho, Rua Dr. Francisco Sa Carneiro, 4400-129, Vila Nova de Gaia, Portugal; Tel +351914314972; Email: jacintarodriguesf@gmail.com

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Convulsions and mental impairment are common. Other associated findings include macrocephaly, microcephaly, hypertelorism, coarse facial features, hemi-hypertrophy, kyphoscoliosis, genital anomalies, inguinal hernia and congenital heart disease [6]. Our patienthad cutaneous, neurologic and musculoskeletal manifestations.

Differential diagnosis includes pigmentary anomalies along the lines of Blaschko such as linear and whorled nevoid hypermelanosis, naevusdepigmentosus and incontinentiapigmenti.

There are no pathognomonic alterations at EEG for hypomelanosis of Ito. Findings of abnormal rhythmic EEG can be an indication of neuronal migration defects [7].

MRI brain findings in patients with HI include hemimegalencephaly, medulloblastoma, cortical malformations, dilated Virchow-Robin spaces, brain atrophy, small discrete bilateral periventricular cysts, abnormal white matter signal, and gray matter heterotopias and other neuronal migration defects [8]. Our patient presented dilated Virchow-Robin spaces at MRI, which is a relatively common abnormality in the HI.

There is no specific treatment, which is based on symptomatology. Care should be multidisciplinary with periodic consultations with a pediatric ophthalmologist, neurologist and orthopedic specialist for this condition. The prognosis is determined by the multisystemic abnormalities [4].

The importance of this case relates to the recognition of the skin typical guiding diagnosis aspect, orienting the study and subsequent follow-up.

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