Sebaceous naevus syndrome with multisystemic manifestations

INTRODUCTION

Sebaceous naevus syndrome, also known as Schimmelpenning-Feuerstein-Mims syndrome,[1] involves the presence of a sebaceous nevus associated with extracutaneous manifestations, mainly neurological, ophthalmological and skeletal. To a lesser extent, it can also affect the cardiovascular, endocrine or urogenital systems.

CASE DESCRIPTION

A male infant born at term, weighing 3850 g. Conceived via in vitro fertilization. Background of polyhydramnios. At birth, presented yellowish plaques on the face (figure 1) and right side of the trunk following Blaschko's lines (figure 2), associated with right ptosis and hemihypertrophy of the right upper limb. MRI of the brain detected a cyst in the left choroid fissure (1.6 cm). Genetic testing on skin sample was positive for HRAS and NOTCH1 genes. At 1 month, the patient developed supraventricular extrasystole, atrial flutter and ventricular tachycardia requiring admission to the pediatric intensive care unit with electrical cardioversion and multiple medications (esmolol, digoxin, flecainide and propranolol), followed by ablation. At 3 months, the patient was diagnosed with asymmetric epileptic spasms, which were treated with everolimus.

DISCUSSION

Sebaceous naevus syndrome[2] is a sporadic, rare disorder caused by postzygotic mosaic mutations in the KRAS, HRAS, NRAS and FGFR2 genes. It presents at birth as yellow-pink plaques with a high association with systemic manifestations,[3 4] especially involving the central nervous system, with a high degree of intellectual disability (80%) and onset of epileptic seizures (57%). Less frequently, it associates with alterations in other systems, such as cardiac involvement, which in our case resulted in high morbidity and a very high risk of mortality.

Management of these patients should be multidisciplinary due to the possibility of potentially serious systemic manifestations.

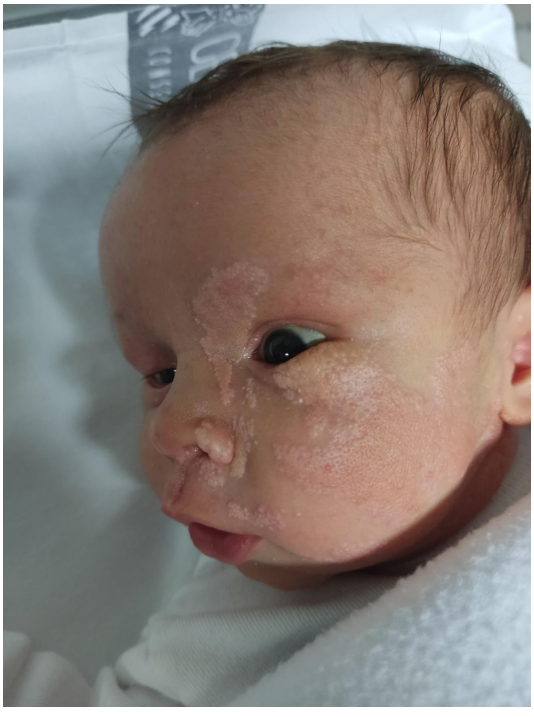

Figure 1 Newborn with yellowish plaques on the face associated with right ptosis.

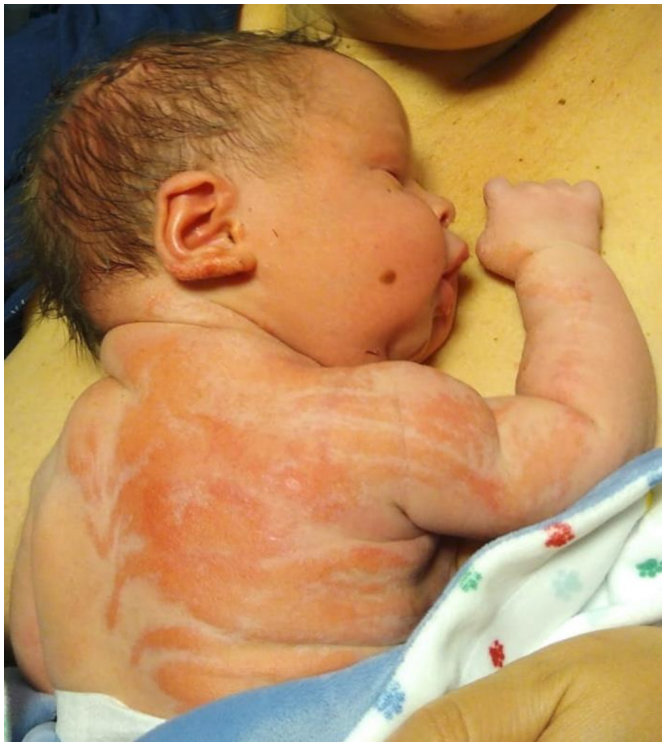

Figure 2 Newborn with yellowish plaques with verrucous appearance on the right side of the trunk following Blaschko's lines.

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