

reveal an infectious trigger. Only a very few instances of hematologic adverse effects such as thrombocytopenia and agranulocytosis have been reported with isotretinoin (4,5). Periodic monitoring of serum triglycerides, cholesterol, and transaminases are recommended but no consensus exists on monitoring other laboratory parameters including complete blood count (1). We believe that the agranulocytosis in this patient may have been due to an idiosyncratic reaction to isotretinoin, but we cannot definitively exclude other cause or a possible coincidental association. Physicians monitoring patients on isotretinoin should be aware of the possibility of this rare, but potentially life-threatening side effect.

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DERMATOLOGIC FEATURES IN PALLISTER–KILLIAN SYNDROME AND THEIR IMPORTANCE TO THE DIAGNOSIS

Pallister–Killian syndrome (PKS) is a dysmorphic condition caused by a tissue limited mosaicism for tetrasomy of 12p. This rare sporadic disorder was first described by Pallister et al in 1977 and later by Killian and Teschler-Nicholas in 1981 (1). Cytogenetic analysis reveals that the majority of fibroblasts of the affected patients have 47

chromosomes, with an extra small metacentric chromosome. We describe a PKS patient with the typical phenotype confirmed by fluorescence in situ hybridization analysis of skin fibroblasts.

CASE REPORT

A 5-month-old boy was referred to the Clinical Genetic Unit of the Paediatric Department because of dysmorphic features and developmental delay. The patient was born at 35 gestational weeks with normal auxologic parameters. After birth, because of the presence of jaundice and dysmorphism in a premature child, a chromosome analysis was performed, which revealed normal karyotype 46,XY.

On examination he had the typical PKS phenotype (Fig. 1A,B) as described in Table 1. Hypo-pigmented cutaneous streaks on the trunk and legs were detected using Wood's light. Cytogenetic molecular examination was performed on skin fibroblasts taken from a hypo-pigmented streak. Fluorescence in situ hybridization analysis detected a supernumerary isochromosome 12p and confirmed the diagnosis of PKS (Fig. 2).

DISCUSSION

Pallister–Killian syndrome is a rare (less than 1:10,000) and sporadic chromosomal abnormality characterized by the mosaic presence of a supernumerary 12p isochromosome. Affected patients have four copies of the short arm of chromosome 12 instead of two and although the anomaly is frequent in fibroblasts, chorionic villi and amniotic fluid samples, it is rarely identified in blood lymphocytes (2). The mechanism by which isochromosomes are formed and the stage at which this occurs is still under debate and not yet understood (3).

The variability in the clinical features and presentation of PKS is great and it is possible to find at one end severe forms with intrauterine death of the fetus and on the other very mild forms. Moreover, recently, instances of oro-facial digital syndrome type IX and of Fryns syndromes have been revealed to be PKS after FISH analysis (4,5). The clinical features that overlap with OFD type IX are digital and oral anomalies, while the clinical features that overlap with Fryns syndrome are coarse face, diaphragmatic hernia, and cleft palate.

The complexity and variability of the clinical phenotype and the presence of overlap features with other syndromes make the molecular cytogenetic test necessary not only for a correct diagnosis, which is essential for genetic counseling, but also for possible prenatal evaluation.

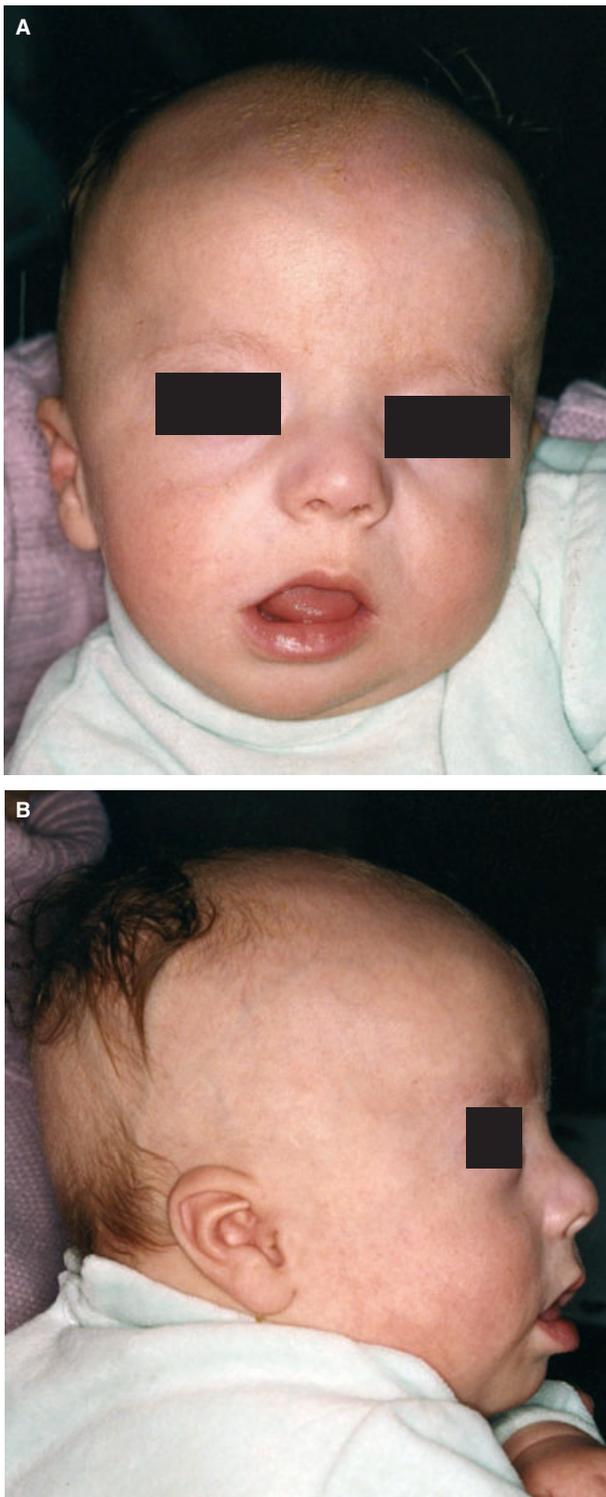


Figure 1. (A) Classical phenotype of Pallister-Killian syndrome in our patient. (B) Supernumerary isochromosome 12p seen on FISH analysis.

TABLE 1. *Cutaneous and Morphologic Features of Pallister-Killian Syndrome*

Streaks of hypo-hyperpigmentation (Wood's lamp)
Prominent forehead
Sparse anterior scalp hair
Abnormally high anterior hairline
Flat occiput
Hypertelorism
Short nose with anteverted nostrils
Short neck
Flat nasal bridge
Long philtrum with thin upper lip
Horizontal palpebral fissures

Various aspects of the phenotype of PKS will interest the dermatologist, such as the facies characterized by an high forehead with sparse anterior scalp hair, abnormally high anterior hairline, sparse eyelashes and hypo-pigmented skin patches, which may be visible only under Wood's lamp. Hypo-pigmented patches distributed along the Blaschko lines are present in about 40% of the patients with PKS and in these sites the rate of fibroblasts owing to the supernumerary 12p is higher (4). Therefore in patients with dysmorphic features a complete dermatologic evaluation for evidence of pigmentation disorders must be performed.

In conclusion, a dermatologic examination of the skin by Wood's lamp is advisable when a suggestive phenotype of PKS is present. Indeed, FISH analysis carried out on skin fibroblasts from hypo-pigmented regions can confirm the diagnosis.

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NEVUS SEBACEOUS SYNDROME WITH FACIAL HEMIHYPERTROPHY

Abstract: Nevus sebaceous syndrome is a member of the epidermal nevus syndromes group, and is characterized by extensive nevus sebaceous, seizures, and mental retardation. We present an affected 5-month-old boy who had facial hemi-hypertrophy and recurrent seizures.

The nevus sebaceous syndrome (NSS) is characterized by the presence of a sebaceous nevus and extracutaneous abnormalities, usually involving organs derived from the neuroectoderm. Recently, we cared for a 5-month-old boy with an extensive nevus sebaceous affecting the right side of the scalp, hemi-hypertrophy of the right side of the face, and recurrent seizures. To the best of our knowledge, this is the first report of this syndrome in West Africa.

CASE REPORT

A 5-month-old boy was referred to our hospital because of recurrent seizures which began on day 17 of life. The seizures were initially focal, involving the left side of the body and later became generalized, lasting about 2–3 minutes. He had a normal twin sibling and no history of similar abnormalities in his family.

Examination found a boy weighing 6.19 kg, with a length of 67 cm. His head circumference was 49 cm. He had marked facial asymmetry with hypertrophy of the right half of the face, extending from the inferior orbital margin down to the submandibular area (Fig. 1), features which his mother stated had been present since birth. The right pinna and the right half of the tongue were also enlarged. He had skin colored and hyperpigmented plaques which were



Figure 1. Marked hypertrophy of the right side of the face.



Figure 2. Sebaceous nevus following Blaschko's lines on the right side of the face.

waxy in some areas and pebbly in others, following Blaschko lines on the right side of his face (Fig. 2). A neurologic examination was normal except for a mild head lag. He had bilateral optic atrophy. Cranial computerized tomography scan revealed