

## Blaschkoid melanosis in a child with pallister killian syndrome

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**Abstract:** We report a case of 9 month old girl with Pallister-Killian syndrome (PKS) with blaschkoid melanosis, which is characterised by hypotonia, pigmentary skin anomalies, craniofacial dysmorphism. PKS is caused by extra isochromosome 12p.

**Keywords:** Blaschkoid melanosis, Pallister-Killian syndrome.

**Keymessage:** Infants presenting with blaschkoid hypermelanosis and typical facial features should be thoroughly examined for PKS

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### I. Introduction

The introduction PKS is a rare chromosomal disorder caused by isochromosome 12p. Isochromosome is a chromosome with two identical arms .ie. either two long arms (p) or two short arms (q). PKS patients have the two usual copies of chromosome 12 and some cells also have the isochromosome 12p, showing tissue limited variable somatic mosaicism<sup>[1]</sup> (presence of two or more populations of cells with different genotypes in an individual who has developed from a single fertilized egg). Isochromosome originates through maternal meiosis II non-disjunction, followed by a rearrangement or centromeric misdivision of prezygotic origin or postzygotic origin<sup>[2]</sup>. Increased maternal age affect has been observed<sup>[1]</sup>. More than 100 cases have been reported in the literature, with prevalence of 1/20,000<sup>[1]</sup>.

### II. Case Report

A 9 month old hindu female was referred to dermatology department for skin biopsy. Patient had linear streaks of hyperpigmentation which appeared to follow lines of blaschko and were distributed all over body asymmetrically. She was born to non-consanguineous parents via c-section and had placental cord around her neck. Birth history was insignificant. She had no difficulty in sucking and swallowing. There was no neurological or respiratory complaint. However she had not attained neck holding till that age. Her social milestones appeared to be poor and she was poor responder to the sound. On examination prominent forehead with frontal and temporal reduced density of hair with abnormal distribution of fat around eyes could be seen [image 1]. Streaks of hyperpigmentation following lines of blaschko were present [image 2 a,b,c,d]. She had bilateral upslanting eyes with hypertelorism and had sparse eyebrows . Nasal root was deep with short nose. Cheeks were chubby. Angles of mouth were downturned [image 1]. Palate was high arched. Right ear appeared to be more prominent than left ear. She had short neck with inverted nipples. Hands were short and stubby with puffy feet. The patient also had naevus of Ito on her right shoulder [ image 3]. Hypotonia was present in lower limbs. Anus was more anteriorly placed. Hair density in the frontal region was reduced [image 4]. Cardiovascular examination was within normal limits. Sonography report of brain revealed right lateral ventricular choroid plexus cyst.

### III. Discussion

Clinically PKS presents coarse facial features<sup>[3]</sup> such as high forehead, hypertelorism, broad nasal bridge, sparse scalp hair mainly in fronto-parietal region, long philtrum or invasion of the upper lip vermilion by philtral skin (Pallister lip, present in most of the patients), sparse eyebrows, depressed nasal bridge, large mandible, bifid uvula, short neck. Facial features are helpful in diagnosis and were consistent with this case. Low muscle tone, streaks of hypo- and hyperpigmentation, profound mental retardation, congenital heart defects, and hernias of the diaphragm are other key symptoms, but no single malformation is pathognomonic of the syndrome.

Facial features along with streaks of hyperpigmentation and hypotonia helped us in diagnosing the case.

Neurologic manifestations such as structural brain malformations and epilepsy may be present, choroidal plexus cyst was present in our case which could also be an incidental finding.

Structural cardiac defects include atrial and ventricular septal defects; pulmonary involvement, most commonly lung hypoplasia secondary to a diaphragmatic hernia; gastrointestinal manifestations may include intestinal malrotation, displacement of the anus, which was also present in this case. Musculoskeletal abnormalities including a growth pattern unique to PKS which consists of an accelerated prenatal growth period followed by a decelerated postnatal growth period [2,3].

75% of patients with PKS have some degree of visual impairment [3]. Congenital diaphragmatic hernia is thought to be very specific of PKS and is frequently a cause of death during the early postnatal period of life. Our patient had typical features along with naevus of Ito which has not previously been reported.

#### **IV. Conclusion**

Pallister killian syndrome is very rarely diagnosed syndrome. Infants need to be completely evaluated to rule out any systemic abnormality so as to provide better life to the patient.

#### **V. Declaration Of Patient Consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

#### **Conflict Of Interest**

Nil

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Nil.

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