SHORT REPORT

Severe developmental delay and multiple strawberry naevi: a new syndrome?

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Abstract

An 18 month old girl with dysmorphic features, severe developmental delay, multiple strawberry naevi, and capillary naevi is described. No previous report of a similar association of features has been identified.

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The patient, an 18 month old girl, is the third child of healthy, unrelated parents with no significant family history. Her two older brothers are healthy. Pregnancy was drug free and uneventful until 32 weeks' gestation when ultrasonography showed an enlarged heart and clenched hands. No specific cardiac anomaly was identified on detailed scanning and placental biopsy showed a normal female karyotype.

Labour started spontaneously at 36 weeks' gestation and the baby was born by vaginal delivery after which she was asphyxiated and required intubation with assisted ventilation. Growth parameters at birth were: weight 2440 g (25th centile) and head circumference 32.7 cm (50th centile). Abnormal features noted at that time included a prominent occiput, tiny anterior fontanelle, narrow palpebral fissures, flexed fingers, and hypotonia.

Early infancy was characterised by developmental delay, feeding problems associated with gastro-oesophageal reflux, and the appearance of strawberry naevi on all parts of the body from the age of 3 weeks onwards. At recent assessment at the age of 16 months, there was severe global developmental delay with no milestones above the three month level. Motor development was particularly delayed with marked central hypotonia. Length and head circumference fell on the 50th centile whereas weight was below the 3rd centile. Notable features included an unusual facial appearance (fig 1) with a high forehead, blepharophimosis, low set, posteriorly rotated ears, thin eyebrows, small nose, high palate, tight oral frenula, and small chin. Iris pigmentation was unusual with concentric layers of different colours. Her hands and feet were long and thin and there was fixed flexion of both index fingers (fig 2). Other findings were a thoracic scoliosis, a prominent capillary naevus on the forehead, and approximately 30 strawberry naevi widely scattered over all parts of the body (fig 3). At the age of 17 months she developed myoclonic epilepsy, although

before the onset of this developmental delay was already severe.

Investigations which gave normal results included routine blood and urine biochemical screen, TORCH screen, repeat karyotype, fundoscopy, echocardiography, and neonatal cranial ultrasonography. Radiography showed thin ribs with no evidence of structural vertebral anomalies. A recent EEG showed hypsarrhythmia.

CT scan of her head showed severe global cerebral atrophy and left sided calvarial bony exostoses, but no evidence of any intracranial haemangiomas.

The most notable clinical features in this patient consisted of severe retardation, with failure to thrive, blepharophimosis, flexed index fingers, and multiple strawberry naevi, findings for which no clear syndromal diagnosis has been forthcoming. Blepharophimosis and camptodactyly occur together in the Marden-Walker syndrome, a poorly defined and possibly heterogeneous disorder which shows autosomal recessive inheritance.^{1,2} Strawberry naevi are not a recognised feature

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Figure 1 Detail to show facial dysmorphism. Note the multiple strawberry naevi and forehead capillary naevi.

of this syndrome in which contractures are usually widespread rather than limited to the index fingers. Blepharophimosis and multiple haemangiomas are recognised features of the fetal alcohol syndrome but this diagnosis is untenable given the lack of exposure to alcohol in utero and the severe degree of global retardation. Haemangiomas and hypsarrhythmia may occur together in the linear sebaceous naevus syndrome, but our patient shows no evidence of the linear pigmented lesions which are the hallmarks of this disorder.³

Widespread haemangiomas occur in disseminated neonatal haemangiomatosis and neurodevelopmental problems may result from intracerebral lesions.⁴ However, dysmorphic features are not associated and our patient did not have any intracerebral lesions on CT scanning.

No published reports of similarly affected

children have been identified, prompting us to suggest that the findings in this child may constitute a 'new' dysmorphic syndrome.

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- 2 Gossage D, Perrin JM, Butler MG. A 26 month old child with Marden-Walker syndrome and pyloric stenosis. *Am J Med Genet* 1987;26:915-19.
- 3 Clancy RJ, Kurtz MB, Baker D, *et al.* Neurologic manifestations of the organoid nevus syndrome. *Arch Neurol* 1985;42:236-40.
- 4 Burke EC, Winkelmann RK, Strickland MK. Disseminated hemangiomatosis: the newborn with central nervous system involvement. *Am J Dis Child* 1964;108:418-24.

Figure 2 Detail of hands to show fixed flexion deformities of the index fingers.

Figure 3 Child aged 16 months, showing multiple strawberry naevi.