

Pallister-Killian syndrome: a common yet under-recognised cause of epileptic spasms

To the Editor,

I read with great interest the recent article by de Palma *et al.* (2012), entitled "Eating-induced epileptic spasms in a boy with MECP2 duplication syndrome: insights into pathogenesis of genetic epilepsies". A common cause of "epileptic spasms" that is often overlooked and under recognised is Pallister-Killian syndrome (PKS).

PKS or "Pallister mosaic aneuploidy syndrome" is a rare cause of epileptic spasms and was first reported in 1977. It is characterised by the triad of learning impairment, craniofacial dysmorphism and, seizures (Yamamoto *et al.*, 2007). Epileptic spasms are seen in 30% of patients with PKS. Nearly 87.5% of all epileptic spasms in patients with PKS first appear before the age of two years (Candee *et al.*, 2012). However, epileptic spasms in patients with PKS may first appear as late as 9.5 years of age. Brain malformations are relatively frequent in PKS patients who develop epileptic spasms (Giordano *et al.*, 2012). Rare variations of the syndrome are seen sometimes. For instance, Chaouachi *et al.* (2010) recently reported a patient with PKS who developed a sacral appendage as well as a cleft palate. Delay in psychomotor development is commonly seen. Craniofacial dysmorphism is typically present. For instance, a prominent forehead and a flat face are usually seen and the nose is small and nostrils anteverted. Oral examination reveals micro-retrognathia and a high arched palate (Cerminara *et al.*, 2010). Ears are typically low-set and dysplastic. Hypertelorism and epicanthal folds accompany the above-mentioned facial changes. Eye examination may also reveal transillumination defects of the iris. The nasal bridge is flat as is the philtrum. Skin examination reveals linear leukoderma. Shah *et al.* (2012), in a recent report, also reported post auricular as well as sacral pits. Examination of the scalp reveals abnormal hair. Delayed ossification of the pubic bones is commonly seen, resulting in skeletal deformities (Sanchez-Carpintero *et al.*, 2005). Various forms of seizures are typically seen in 59% of patients with PKS. Neurological examination usually reveals hypotonia. Gastrointestinal complications, such as diaphragmatic hernias, are fairly common, as are cardiac anomalies. For instance, Jamuar *et al.* (2012) recently reported aortic dilatation in an infant with PKS. A history of foetal macrosomia can usually be elicited.

EEG may reveal modified hypsarrhythmia. Diffuse biphasic slow waves with super-imposed low-voltage

fast activity is usually seen on ictal EEG. Bursts of "slow waves" or "focal spikes" with disorganised background activity are typically seen on interictal EEG during wakefulness (Kostanecka *et al.*, 2012). Fluorescence *in situ* hybridisation typically reveals mosaic tetrasomy 12p. This test is usually performed in cultured fibroblasts. X-rays of the chest may reveal flared anterior ribs. Broad femoral metaphysis is another common radiological finding. Murakami *et al.* (2011) have recently utilised amniocentesis during the third trimester of pregnancy for the antenatal diagnosis of PKS. Increased "nuchal translucency" may be noted on prenatal ultrasound.

Anti-seizure medications should be instituted immediately in PKS patients with "epileptic spasms". Yamamoto *et al.* (2007) have reported success with concurrent administration of valproic acid and high-dose pyridoxine in these patients. Surgery may be needed in a few cases of PKS. For instance, Baglaj *et al.* (2008) recently reported a patient with PKS who developed a midgut volvulus requiring emergency laparotomy.

As is obvious from the above discussion, PKS is a fairly common cause of "epileptic spasms" and should be considered in the differential diagnosis for all patients presenting with this form of seizure. □

Disclosures.

The author has no conflicts of interests to disclose.

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