be instituted to prevent menopausal symptoms after hysterectomy and bilateral oophorectomy.

REFERENCES


Magnetic resonance imaging of the cerebral malformation in Miller-Dieker syndrome

A case report

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Summary

Absent or defective cortical gyri (lissencephaly) combined with a characteristic phenotypic appearance was first reported by Miller and Dieker in 1963 and the clinical, computed tomographic and pathological features of this syndrome have been extensively reviewed. We report on magnetic resonance imaging of the brain in a sporadic case of this syndrome.


Twenty-eight cases of Miller-Dieker syndrome have been reported since its first description in 1963. The minimal diagnostic criteria proposed by Dobyns et al. include facial malformations (micrognathia, abnormal ears), severe postnatal growth retardation, and computed tomographic (CT) or pathological evidence of 'lissencephaly' or defective gyri. Although electro-encephalographic findings and real-time ultrasonographic observations may suggest the presence of cortical dysgenesis, the diagnosis is usually made by the characteristic appearance of the brain on CT. The multiplanar facility and resolution of magnetic resonance imaging (MRI) now makes this the investigation of choice.

Case report

A 6-month-old boy was referred from a rural hospital with failure to thrive. He was one of twins, delivered after a normal, term pregnancy. The parents were unrelated. The other twin and another sibling were developing normally. Feeding problems had been present from birth and contributed to severe growth retardation. There was no history of seizures. On examination the weight, length and head circumference were below the 3rd percentile. The child had severe spastic quadriplegia with multiple joint contractures and bilateral talipes equinovarus. Dysmorphic craniofacial features included an oval face, small low-set ears, high-arched palate, retrogloathia, slightly upturned nares and a broad nasal bridge. There was bilateral camptodactyly of the 4th and 5th fingers with single palmar creases, and the left testis was undescended. CT of the brain revealed smooth hemispheric surfaces, with prominence of the sylvian fissures and moderate dilatation of the posterior horns of both lateral ventricles. Axial T2 MR images (spin-echo technique) were obtained using a 0.5T scanner. These showed coarse convolutions anteriorly, underdeveloped temporal opercula and complete agyria of the occipital lobes (Fig. 1). The superficial grey matter was widened occipitally, with abnormal configuration and high T2 signal intensity of central white matter.

No chromosomal studies were done.
Fig. 1. T2 MR images. Left: late echo (TE 27); and right: early echo (TE 120). These demonstrate increased thickness of grey matter with coarse gyri frontally and smooth occipital and temporal lobes. The sylvian fissures and posterior cortex are widened.

Discussion

Agyria or lissencephaly implies a smooth hemispheric surface due to a disorder of early neuronal migration. More commonly, the cortex is not uniformly smooth, showing areas of primitive sulcation, with broad, shallow gyri referred to as pachygyria. Infants with this condition often have marked hypotonia at birth which progresses to severe spasticity. Agyria-pachygyria may occur as an isolated developmental defect, but is also found in a number of other syndromes, such as Miller-Dieker, which was diagnosed clinically in our patient.

CT in these cases reveals a smooth cerebral surface and absent opercula, accounting for the characteristic 'figure-of-eight' appearance of the brain.

The white/grey matter interface may also appear abnormal. The relatively poor resolution and uniplanar images of CT make it likely that many cases diagnosed in the past as lissencephaly, probably belong to the partial agyria-pachygyria group of anomalies. While CT in our patient was suggestive of CNS malformation of the lissencephalic type, MRI permitted a more precise diagnosis of partial agyria, reflecting a milder form of the developmental defect. The superior anatomical detail afforded by this imaging modality has recently been reported by others, and MRI is now regarded as the optimum investigation in all cases of CNS malformation.

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