

DESCRIPTION

We present a 28-year-old right-handed male who, at 17 weeks' gestation, suffered a traumatic amniocentesis, and was subsequently born with extensive right-sided cerebral, cerebellar and brainstem damage. The initial amniocentesis was abandoned and repeated 2 weeks later, with normal results, subsequent prenatal ultrasound scans were normal. The patient had an uncomplicated delivery at term, however a complete right sided ptosis and marks on the right parietal region of the scalp were noted. By 4 months of age, a right sided third nerve palsy, a left sided hemiplegia and left visual field defect had been identified, and infantile spasms developed, which were treated successfully with ACTH. No further seizures occurred until habitual seizures began at 7 years of age, consisting of focal seizures with, and without, loss of awareness, with blank spells, abdominal discomfort, head turning, agitation and disorientation. Although initially controlled with antiseizure medication, seizures became refractory within 3 years, occurring several times per day despite extensive medications and vagal nerve stimulation. In recent months, atonic seizures developed, characterised by sudden weakness of the lower limbs causing collapse, not associated with loss of consciousness, with risk of injury and a significant effect on his quality of life. There have not been secondarily generalized tonic-clonic seizures. He achieved undergraduate level education and can walk despite a left hemiparesis.

MRI shows extensive cortical encephalomalacia of the right frontal, temporal and parietal lobes, including an atrophic hemicranium, clefts from the cerebral surface to the dilated right lateral ventricle, , and a small, dysplastic right cerebellum and brainstem (Fig 1 and 2). There is more extensive damage than typically occurs with schizencephaly. The left side of the brain is normal. Scalp video-EEG telemetry shows multifocal areas of seizure onset and interictal epileptic activity in the right hemisphere. At present, a functional right hemispherectomy is under consideration as a treatment for the epilepsy.

Schizencephaly is a rare congenital malformation of the brain, with a prevalence of approximately 1.48/100,000 live births in the UK¹, and often presents with severe and life-long neurological sequelae including motor and cognitive dysfunction, and epilepsy². Whilst the precise aetiology is unknown, insults in early foetal brain development are implicated, including CMV infection, genetic factors, and maternal trauma. Traumatic amniocentesis has, in rare circumstances, been documented as a cause of foetal brain injury³, including schizencephaly⁴. The extent of the damage is widespread, and has a similar pattern of cerebral, brainstem and cerebellar hypoplasia and atrophy to other documented cases of damage secondary to traumatic amniocentesis⁴.

LEARNING POINTS

- Amniocentesis may result in severe brain injury, resulting in a fixed deficit and drug-refractory epilepsy.
- Amniocentesis-induced foetal injury is extremely rare, and the risk may be minimised with the use of continuous ultrasound guidance.

REFERENCES

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FIGURE LEGEND

Fig 1: Coronal FLAIR MRI demonstrating, right hemispheric encephalomalacia, atrophy, particularly of the right temporal lobe, cleft and hypoplastic right cerebellum and brainstem .

Fig 2: T1 axial MRI demonstrating extensive damage to the right temporal and occipital lobes.