

Bilateral periventricular nodular heterotopia – neonatal cranial ultrasound

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A female neonate was delivered at 30 weeks' gestation to a mother with suspected bilateral periventricular nodular heterotopia (PNH) on computed tomography brain scan. Antenatal ultrasound (US) performed at 28 weeks' gestation showed mild lateral cerebral ventricle dilatation, but no other fetal anomalies. Clinical examination revealed mild dysmorphic features: facial and skull asymmetry, square-shaped ears, and long tapering fingers. Chromosomal analysis showed a 46XX karyotype.

Cerebral US revealed asymmetry and irregular dilatation of the lateral ventricles (Figs 1 and 2). These findings are nonspecific and can be associated with post-infectious, post-ischaemic and/or post-haemorrhagic periventricular infarction.¹ However, a scan through the extreme lateral margin of the lateral ventricle revealed multiple large nodules covering the lateral aspects of the frontal horn and body of the lateral

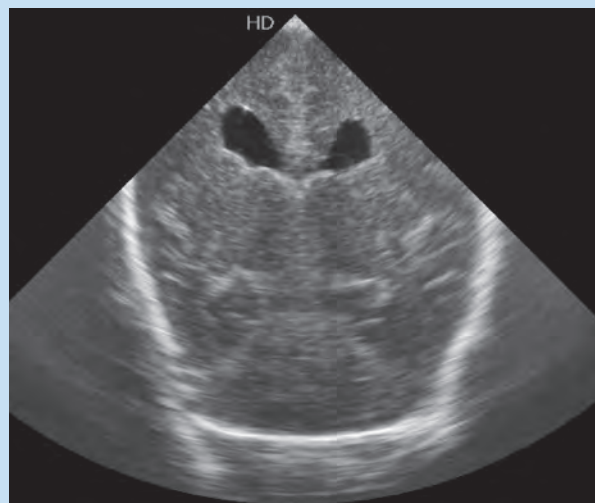


Fig. 2. Coronal US scan through the body of the lateral ventricles.

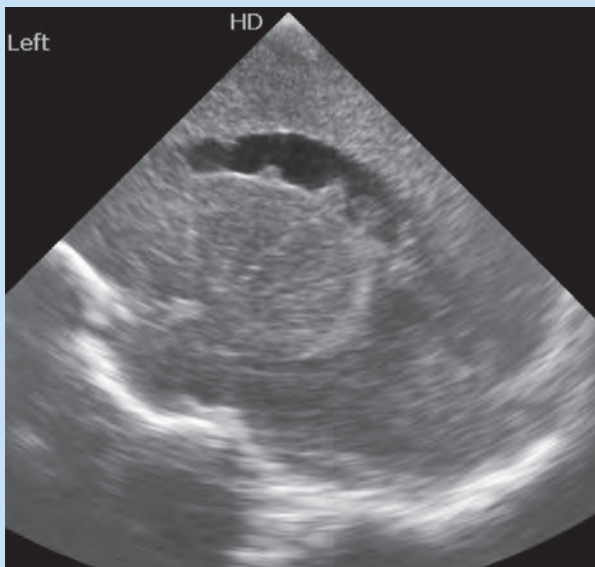


Fig. 3. Parasagittal US scan through the lateral aspect of the left lateral ventricle.

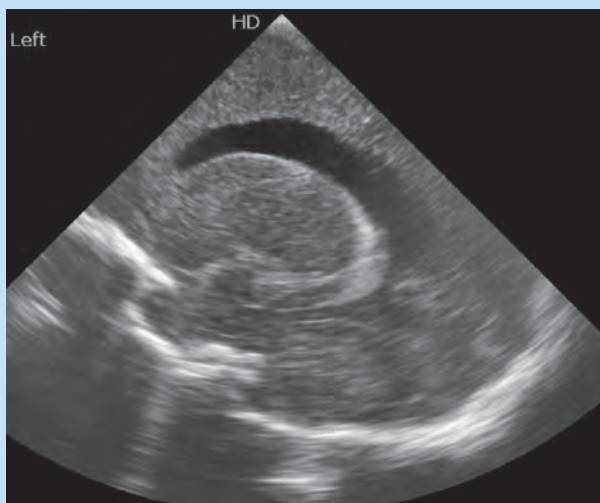


Fig. 1. Parasagittal US scan through the body of the left lateral ventricle.

ventricles (Fig. 3). The nodules were better demonstrated by rotating the US probe to focus predominantly on the lateral and inferior aspects of the frontal horn (Fig. 4).

PNH is a neuronal migration disorder characterised by nodules of ectopic neurons adjacent to the lateral ventricles, owing to failed migration of neurons from the germinal zone towards the cortical plate.² Classical bilateral PNH is the most common type and, in more

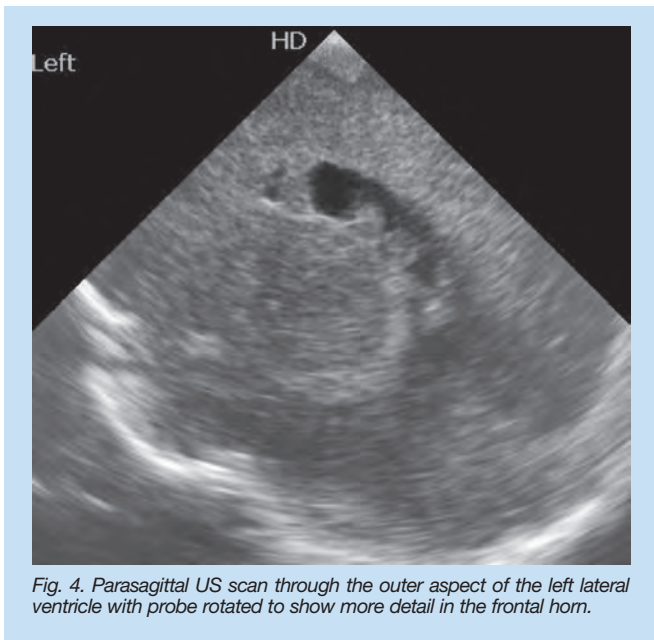


Fig. 4. Parasagittal US scan through the outer aspect of the left lateral ventricle with probe rotated to show more detail in the frontal horn.

than 50% of cases, is due to mutations of the X-linked *Filamin A* gene.² Most affected females develop seizures, with variable age of onset and type. Dyslexia is common but intellect is usually normal to borderline. Hypoplasia of the cerebellar vermis and valvular heart disease are commonly associated, but neither were present in this neonate.³

PNH is usually diagnosed on magnetic resonance imaging but the US scans obtained in this patient demonstrate nodules that are typical of bilateral PNH.¹ Previously published ultrasound images of neonates with this condition⁴ have not clearly demonstrated the distribution and size of the nodules as shown in this neonate.

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