DESCRIPTION
A baby boy was born to healthy, unrelated parents at 34 weeks gestation by caesarean section in view of an antenatal ultrasound diagnosis of hydrocephalus. Mother had a previous miscarriage at 13 weeks gestation of a twin pregnancy. At birth, the boy weighed 2.47 kg, was well and did not require resuscitation. Pregnancy was unremarkable with no significant events. At birth, a large head circumference was noted, but fontanelles were soft. The Apgar score was 9 at 1 and 10 min, baby had a good cry and was moving all four limbs. A CT scan of brain was done at 1-day old which showed obstructive hydrocephalus at the level of the cerebral aqueduct. A ventriculoperitoneal (VP) shunt was inserted at 1-week old. The boy showed slow development over the subsequent years, he never suffered from seizures, however required growth hormone therapy. A small atrial septal defect was noted on echocardiogram. An MRI scan of head at 8 years and 8 months was done in view of developmental delay. This showed a normally sited VP shunt and no hydrocephalus.

Rhomboencephalosynapsis was diagnosed with the absence of the cerebellar vermis and fusion of the cerebellar hemispheres, with transversely oriented inferior cerebellar folia. To date, no genetic diagnosis has been found.

Learning points
- Rhombencephalosynapsis is a rare, idiopathic abnormality of the posterior fossa, characterised by the absence of the cerebellar vermis and fusion of the cerebellar hemispheres. Fusion may or may not include the cerebellar peduncles and colliculi and the dentate nuclei.
- Associated with several other anomalies, particularly of the midline. These include supratentorial anomalies, most commonly ventriculomegaly due to aqueductal stenosis. The corpus callosum may be normal, dysplastic or hypoplastic.
- Cerebellar abnormalities may be difficult to be detected on CT; MRI outlines the abnormality more clearly and hence rhombencephalosynapsis has been increasingly reported since the invention of MRI.

Contributors
SA wrote the attached case report, under the guidance of RG, who provided the images obtained during routine practice.

Competing interests
None declared.

Patient consent
Guardian consent obtained.

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REFERENCES