7q11.23-q21.2 Microdeletion is associated with moderate structural brain abnormalities and global developmental delay: first report

George T Vasileiadis,¹ Jennifer Carter,² Richard H Scott²

¹Department of Neonatology, BHR University Hospitals NHS Trust, London, UK

²Department of Clinical Genetics, North East Thames Genetic Service, London, UK

Correspondence to

Dr George T Vasileiadis,
George.Vasileiadis@
bhrhospitals.nhs.uk

Accepted 9 September 2015

DESCRIPTION

We present a case of a male infant born at 29⁺⁶ weeks (weight 1670 g) following an antepartum haemorrhage. Antenatal scans did not reveal any abnormality; the infant had a high arch palate and

small germinal matrix bleed but no intraventricular haemorrhage. He was noticed to be moderately hypotonic at the age of term equivalent. Brain MRI revealed moderate ventriculomegaly bilaterally with no hydrocephalus (figure 1), and moderate hypoplasia of the corpus callosum (figure 2).¹ Interestingly, detailed developmental assessment (Bayley III) at the adjusted for prematurity age of 13.5 months showed a global developmental delay affecting cognitive, communication and motor domains (percentile ranks: 1, 4, 0.1, respectively).² His weight plotted on the 91st centile and his head circumference on the 50th.

Genetic studies: microarray detected a heterozygous 16.7 Mb microdeletion at chromosome 7q11.23-q21.2. Fluorescence in situ hybridisation analysis using probe RP11-185D15 confirmed the analysis. The deletion was not present in either parent, encompassed 76 known genes and did not overlap the Williams-Beuren critical region. This is the first report of such deletion and further cases with closely overlapping microdeletions are required to clarify which of the genes in the deleted interval contribute to the child's phenotype and long-term outcome.

Figure 1 Enlarged frontal and occipital horns of both lateral ventricles (arrows).

Figure 2 Hypoplastic corpus callosum (between arrows).

To cite: Vasileiadis GT, Carter J, Scott RH. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2015-212175

Learning points

- ▶ Differential or overlapping diagnoses: Prematurity is associated with ventriculomegaly, and both may be associated with hypotonia and developmental delay. However, genetic abnormalities can also be associated with hypotonia and developmental delay.
- ▶ Callosal abnormalities are present in a significant proportion of infants with ventriculomegaly, irrespective of prematurity.
- ▶ Combined callosal abnormality/ventriculomegaly and global developmental delay may be associated with 7q11.23-q21.2 microdeletion.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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