Unusual neuroimaging features in a patient with chromosome 11q14.1–11q23.2 deletion

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DESCRIPTION
An early adolescent patient with a history of chromosome 11q14.1–11q23.2 deletion presented for the evaluation of new onset seizures. Semiology was suggestive of a generalised tonic-clonic seizure without focal onset. Electroencephalogram showed no definite epileptiform abnormalities, focal slowing or electroclinical seizures. Examination was remarkable for baseline cognitive impairment, microsomia, micrognathia, a short neck and low-set posteriorly rotated ears. MRI revealed an unusual pattern of prominent pial enhancement (figure 1A–D) that corresponded to dilated tortuous vessels on T2-weighted sequences (figure 1E–G). The demonstration of superficial cerebral vein tortuosity contrasts with the pattern of leptomeningeal enhancement that may present in neoplastic or infectious neurological diseases.1 The brain parenchyma was of normal signal intensity on all conventional MRI sequences without focal abnormalities.

Interstitial deletions of the long arm of chromosome 11, spanning from bands 11q13 to 11q23, are rarely observed and reported.2 Clinical manifestations that may present with interstitial 11q deletions are palate anomalies, developmental delay, dysmorphic features, and in some cases, congenital heart malformations, hypotonia, seizures and renal abnormalities.2 3 To our knowledge, only six interstitial 11q deletion cases report seizures, and seizures have been correlated with the more distal 11q22–11q23 region.2 The deleted region in our patient is approximately 33 Mb in size and contains 97 Online Mendelian Inheritance in Man annotated genes that contribute to diverse cellular functions. Genetic mechanisms resulting from interstitial 11q deletions may underlie vascular abnormalities and seizure development, however, specific genotype-phenotype correlations are yet to be discovered.2–4 We hypothesise that the angiomotin-like protein 1 gene located on chromosome 11q21 which is involved in the regulation of endothelial cell migration and angiogenesis may be in part responsible for the observed unusual MRI findings.4 5 Epilepsy may arise from vascular mechanisms such as neurovascular coupling or blood flow dysregulation.6 Our case is the first to highlight intracranial vascular findings in a patient with this rare interstitial 11q deletion. MRI evaluations in other interstitial 11q deletions have commonly demonstrated normal findings,2 3 7–9 with only a few cases revealing white matter abnormalities, mild cerebral atrophy and corpus callosum hypoplasia.10 11 In our case, narrowing of the foramen magnum may represent a possible perpetuating factor of blood flow dysregulation and subsequent vascular congestion. Underlying cerebrovascular disorders, such as moyamoya disease, are more unlikely in our patient as specific MRI findings, including diffuse leptomeningeal enhancement and increased deep medullary vein visibility, are often associated.12 Sturge-Weber syndrome, a congenital vascular malformation involving the brain, was

Figure 1 Atypical MRI features in a patient with 11q14.1–11q23.2 deletion. Post gadolinium MRI sequences (A–D) revealed prominent leptomeningeal enhancement (arrows) without other abnormalities. T2-weighted sequences (E–G) demonstrated enlarged, tortuous pial vessels (arrows) corresponding to areas of leptomeningeal enhancement without evidence of other intracranial abnormalities.
also considered. However, there were no indicative neurocutaneous markers or imaging findings, such as leptomeningeal angiomatosis with dilation of the choroid plexus. Additionally, intracranial dural arteriovenous fistulas (DAVF) were considered in the differential diagnosis of vessel tortuosity and congestion, however, there was no evidence of a pseudophlebitic pattern on imaging. Associated findings with the presence of a pseudophlebitic pattern, such as cerebral oedema, chronic haemosiderin deposition and dilated transmedullary veins, were also not demonstrated. Therefore, an underlying DAVF is a less likely aetiology of intracranial vessel tortuosity in our case.

Overall, our case highlights the clinical phenotypes that may arise from a chromosome 11q14.11–11q23.2 deletion while demonstrating the first intracranial vascular findings in the MRI evaluation of an interstitial 11q deletion case.

**Learning points**

- Prominent leptomeningeal enhancement and dilated, tortuous pial vessels may appear in the neuroimaging evaluation of rare interstitial 11q deletion cases unrelated to underlying cerebrovascular disorders, dural arteriovenous fistulas, or infectious and neoplastic processes.
- MRI evaluations in interstitial 11q deletion cases commonly demonstrate normal findings, with the exception of a few reports in the literature highlighting white matter abnormalities, mild cerebral atrophy and corpus callosum hypoplasia.

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**REFERENCES**


