A 6 year old girl followed for severe mental and neuromotor retardation was examined for genetic abnormalities. The daughter of non-consanguinous parents, and sister of a normal brother, the child had important visual problems and significant facial dysmorphism. Her cranial MRI showed mesial temporal sclerosis with hypoplastic sulci dilation, hypotrophy and cornual deformity. She had a cytogenomic microarray analysis performed. Her results showed a loss involving chromosome 1p13.3p13.2 (7.32 Mb), indicating a deletion in this region, region that is held important for cases that were evaluated for neurological impairment, congenital anomalies and facial dysmorphism. The cytogenomic microarray analysis also showed a loss involving chromosome 2q23.3q24.1 (4.04 Mb), indicating a deletion in this region. A de novo deletion overlapping this region has been reported to be disease causing in one patient (Malbin J et al), a deletion that has been classified as Variant of Uncertain Significance.

RapidSureDeepDive is a cytogenetic method which detects copy number variants with a much higher resolution than conventional cytogenetic analysis. This technology, which does not require staining or cell culture, uses specially designed chips to study the DNA from various sample types. The deletions found in our patient are quite rare, and maybe the variant of the second deletion will be reclassified as disease causing if proved to be denovo.

A 6 year old girl followed for severe mental and neuromotor retardation was examined for genetic abnormalities. The daughter of non-consanguinous parents, the child was found to have mesial temporal sclerosis with hypoplastic sulci dilation.

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Conclusion WBS is rare disorder characterised by wide range of symptoms and signs. Clinical diagnostic criteria are available for WBS, but confirmation of diagnosis requires detection of microdeletion on chromosome 7q11.2. Early recognition of specific patterns of the disorder, such as facial dysmorphic features together with heart abnormalities, cognitive deficiency and vision problems, is crucial because those patients need to take speech therapist and psychologist therapy as soon as possible because it improves their integration in social environment. Management is focused on treatment of symptoms (eg. hypertension, hypercalcaemia), psychological and psychiatric evaluation and speech therapy.