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, hypothyrophy 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. The deleted region does not include MBD5, a gene commonly implicated in intellectual disability and hence the deletion has been classified as Variant of Uncertain Significance. The variant will be reclassified as disease causing if proved to be denovo.

We present the case of three patients, two girls at the ages of one and three years and a boy at the age of two years. All three patients have healthy parents and brothers and sisters without known chronic or genetic diseases. All three were presented with facial dysmorphic features which include prominent ears, palmar crease on left palm, pulmonary stenosis, septal defect, gastroesophageal reflex disease (GERD), hypothyroidism and endocrine disorders; hypercalcemia, hypercalciuria, hypothyroidism and early puberty. The male patient also has aortic stenosis, while absence of NCF1 gene is related to hypertension. Patients with WBS are characterized by cardiovascular diseases, facial dysmorphic features, intellectual disability, unique personality character and endocrine abnormalities.

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, hypercalcemia), psychological and psychiatric evaluation and speech therapy.