Introduction The effect of this deletion has an impact on development of various organs and glands. 16 p13.3 microdeletion is a rare genetic condition with a variable phenotype spectrum. RBFOX1 [RNA binding protein] located on chromosome 16 p13.3, is one of the three members in Fox gene family encoding splicing factors. We would like to present this case report to elucidate the association of this alteration with hypothyroidism and epileptic seizures as only one case of this association has been reported in the literature.

Case report An 8 year old boy presented with recurrent afebrile, unprovoked seizures and behaviour difficulties. EEG was supportive for multifocal epilepsy. The MRI brain scan was normal. Blood results showed persistently high TSH levels and low free T4. Subsequent workup showed a high TPO level and the endocrine team made a diagnosis of autoimmune hypothyroidism. Genetics investigations showed that the patient had a 16 p13.3 microdeletion. A similar genotype was noted in the mother though she was phenotypically normal.

Discussion RBFOX1 is an important splicing factor identified as a binding protein of ATXN2, suggesting a role in neurologic function. It is highly expressed in the brain, skeletal muscle, heart, other organs and glands. Any alteration with this specific locus is associated with variable phenotypes affecting various body systems. These include mental retardation, epilepsy, mental health disorders, congenital heart defects, obesity and diabetes along with other endocrine problems. The introduction of array comparative genomic hybridization (CGH) has provided the ability to map DNA copy number variations (CNVs) genome with high resolution. Only one patient with this CNV has been reported to have congenital hypothyroidism and associated epilepsy. In our patient though the CGH array has helped in mapping out the genetic defect however its role in hypothyroidism is yet to be established.

Conclusion Patients presenting with multiple system involvement, especially involving the endocrine system with associated developmental delay and neurobehavioral and neuropsychiatric problems should undergo CGH array analysis as part of their endocrine assessment. Moreover endocrinologists need to be aware of this copy number variation as it may have a huge impact on future pregnancies of the patient.

Febrile convulsions (FC) are seizures in children under five associated with fever. They are reported in 2%–5% of children with good prognosis. However, the risk of epilepsy may be increased. This study reported the first Scottish cohort and observed current clinical practice, outcome and associations with socioeconomic status. A FC database was created using routinely collected clinical data of all children attending A and E. A Scottish prevalence of 390/100,000 children/year was elucidated with 2.6% of children experiencing FCs by their fifth birthday. Children from deprived areas were overrepresented. It is established that only very young children or children with complex convulsions should be admitted and further investigations are usually unnecessary. However, 72.6% of children were admitted and 10.4% received an EEG which is inappropriately high. Epilepsy was diagnosed in 2.6% of children, an increase from the 1% population risk. However, many of these children had other abnormalities. Only 1.5% of otherwise healthy children developed epilepsy. Overall, the danger of FCs should not be overstated as they only slightly increase the risk of epilepsy in an otherwise healthy child. This should be explained to quell parental anxieties and reduce unnecessary admissions and investigations.