Further investigations including genetic analysis revealed a mutation in NFKB2 protein (heterozygous c.2557>T). Parental blood samples too were taken for genetic studies. They are non-consanguineous.

The diagnosis was mutation in NFKB2 leading to Common Variable Immunodeficiency (CVID) phenotype associated with hypogammaglobulinemia, alopecia and onycodystrophy. Because of close relationship with endocrinopathies he was referred to endocrine team.

He is on prophylactic azithromycin. Protopic ointment 0.1% applied topically to scalp and nails. Immunology team suggested that he will need immunoglobulin replacement therapy in the future. He may require oral immunosuppressents if the response to the recommended therapy is inadequate.

Discussion Our patient with alopecia, nail dystrophy and hypogammaglobulinemia has genetic abnormality with mutation NFKB2 protein. There were previous reported patients carrying NFKB2 mutation demonstrated endocrinopathies in addition to ectodermal abnormalities together with other autoimmune disorders such as vitiligo and autoimmune thrombocytopenic purpura. Therefore the patient we reported also has been directed to rule out these endocrinopathies especially adrenocortical deficiencies and autoimmune disorders.

Conclusion Patients with alopecia, ectodermal dysplasia and hypogammaglobulinemia has genetic abnormality with mutation NFKB2 protein. There were previous reported patients carrying NFKB2 mutation demonstrated endocrinopathies and in addition to ectodermal abnormalities together with other autoimmune disorders such as hypogammaglobulinemia, genetic abnormalities (CVID) and it’s links with endocrinopathies and auto immune disorders.

Aims Moyamoya is a rare progressive occlusive cerebrovascular disease, affecting both children and adults in a bimodal age pattern. The peak incidence in children is 7–10 years old and can present with a variety of clinical scenarios.

Our aim is to describe the clinical presentation, management and outcome to date of a now 5 year old boy who was diagnosed with Moyanoya at 22 months of age.

Methods We report the presenting features, examinations findings and results of radiological images and laboratory investigations, treatment and natural history with regard to our now school age patient.

Results A 22 month old boy presented to the ED following a fall to one side and an episode of high pitched crying. He was afebrile with no recent temperatures or illnesses. His history was notable for a previous afebrile seizure at five months of age lasting over one hour, and recent concerns regarding developmental delay. He had previously had a normal MRI and CT brain, and a normal EEG.

The seizure was eventually controlled on phenoobarbione, and an urgent CT brain showed frontal lobe ischaemia with chronic infarct in the left parietal lobe. He was transferred to Temple St Hospital and subsequently diagnosed with moyamoya. He attended Great Ormond Street Children’s Hospital for several revascularisation surgeries.

He was diagnosed with autism and a sensory processing disorder, and is fed via a gastrostomy tube.

He is now five years old, and has been seizure free for several years. He is ambulant, with over 100 words and good comprehension.

Conclusions Our case highlights the success of multidisciplinary input, sophisticated sub-specialist care, international expertise and devoted parenting in contributing to the quality of life, despite this devastating disease, in our young patient to date.