Further investigations including genetic analysis revealed mutation in NFKB2 protein (heterozygous c.2557>T). Parenteral 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 hypogammaglobulinaemia, alopecia and onycho dystrophy. 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 immunosuppressants if the response to the recommended therapy is inadequate.

Discussion Our patient with alopecia, nail dystrophy and hypogammaglobulinaemia 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 auto immune thyroidopathy. 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 hypogammaglobulinaemia has genetic abnormality with mutation NFKB2 protein. Variable Immunodeficiency (CVID) phenotype associated with hypogammaglobulinaemia, alopecia and onycodystrophy.

P109 AN UNUSUAL CASE OF AN ANTENATAL DIAGNOSIS OF HUGE NECK MASS; A THYROID IMMATURE TERATOMA IN A NEWBORN

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Introduction Head neck teratomas are rare benign tumors consisting of 3–5% of all teratomas (1,2). Teratomas are embryonal neoplasms that arise when totipotent germ cells escape the developmental control of primary organizers and give rise to more or less organoid masses in which tissues derived from all three blastodermic layers (ectoderm, endoderm, and mesoderm) can be identified. Their histologic features are therefore heterogeneous and may include cystic or solid areas with organoid patterns as well as mature or immature components.

Case description We report a case of an antenatal diagnosis of a huge neck mass on antenatal ultrasound and fetal in utero MRI at 33 weeks gestation. He was born by ELSCS at 38 weeks gestation and an endocochlear intubation was performed directly to maintain airway. The MRI neck postnatal confirmed a well circumscribed multicystic and solid anterior neck mass 9.7x5.2x4.9 cm. Surgical resection of the mass and confirmed a well circumscribed multicystic and solid anterior neck mass and development.

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 phenobarbitone, 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.

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.

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.

Discussion Cervical teratomas in children are almost always benign but locally are aggressive. They can present with respiratory airway compromise and excision is required. A multidisciplinary management is needed with multiple specialties involvement. If complete thyroid tissue is removed replacement therapy is required post operatively. Risk of hypocalcemia secondary to hypoparathyroidism developing post operatively should be monitored and treated accordingly.

REFERENCES
