type of muscular dystrophy, with an incidence of 1 in 3500 male births. The initial feature in most boys with DMD is a gait disturbance, with an onset often before age 3 years. Although there is not curable, it is treatable with an importance of an early diagnosis.

**Case presentation** A 26 months old boy presented to our Paediatric Assessment Unit with a history of fever up to 38°C, vomiting, and poor oral intake. His urine was red in colour, but dipstick and microscopy negative for blood. He had no significant past medical history. Vaccinations were up to date. No history of foreign travel. His development was normal.

On examination he had signs of an upper respiratory tract infection. His inflammatory markers were elevated and he was commenced on IV Augmentin. He received IV fluids antipyretics and anti-emetics.

The following day he refused to weight bear. On repeat blood tests, his ALT value was elevated and remained high on repeat testing. Ancillary test for assessment of liver disease were requested. A Creatine Kinase (CK) was requested and showed a value of 20248 IU/L.

He started to weight bear after 4 days and this continued to improve. CK values decreased quickly in the following two weeks. His neurological exam was normal at clinic follow up 2 months post discharge and his LFTs have now returned to normal.

**Discussions** In the absence of liver pathology, raised transaminase may be an early sign of occult myopathy and such patients should have CK levels checked to look for evidence of muscular involvement as happens in polymyositis and muscular dystrophy as in Duchenne’s (DMD).

In DMD, CK levels typically rise above 10,000 IU/L, but they do not return to normal in a few weeks time. It helps to make a clinical differential diagnosis without performing complex genetic tests for Duchenne’s. Laboratory investigations together with the coryzal symptoms from the onset were consistent with our diagnosis of acute viral myositis.

**P108 A RARE CASE REPORT WITH ALOPECIA, ONYCODYSTROPHY AND HYPOGAMMAGLOBULINAEMIA - COMMON VARIABLE IMMUNODEFICIENCY (CVID)**

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**Introduction** Common Variable Immunodeficiency (CVID) is a condition characterised by primary hypogammaglobulinaemia and in consequence increased susceptibility to infections and less frequently ectodermal abnormalities. Additional endocrinopathies have been identified with hypoadrenalism, growth hormone deficiencies etc in the previously reported patients.

**Case presentation** A 2 and half year old boy presented to paediatric outpatient of Wexford General Hospital with a history of alopecia, onychodystrophy and recurrent infections. Alopecia gradually got worse within one year. He was treated for chest and ear infections with several courses of antibiotics.

On examination we found that his growth was satisfactory. Apart from alopecia and nail dystrophy there were no other abnormalities noted.

Investigations revealed normal full blood count and electrolytes. Low immunoglobulins in all domains were the striking abnormality. Lymphocyte subset and thyroid screening were normal. He was referred to the immunology team.
Further investigations including genetic analysis revealed mutation in NFKB2 protein (heterozygous c.2537>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 immunosuppressants 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 auto immune thrombocytopenic purpura. Therefore the patient we reported also has been directed to rule out these endocrinopathies especially adenocortical deficiencies and autoimmune disorders.

Conclusion Patients with alopecia, ectodermal dysplasia should be investigated and monitored thoroughly in order to confirm or exclude the involvement with other disorders such as hypogammaglobulinemia, genetic abnormalities (CVID) and its links with endocrinopathies and autoimmune disorders.

REFERENCES

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 Moya Moya 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 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.

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.