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.

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THUMB AUTO-AMPUTATION FOLLOWING TOURNIQUET SYNDROME

¹Robert McGrath*, ¹Laura McCarron, ²Kevin Cahill, ²Patricia Eadie, ¹Sinead Harty.
¹Department of General Paediatrics, Our Lady's Children's Hospital, Crumlin, Dublin, Ireland;
²Department of Plastic Surgery, Our Lady's Children's Hospital Crumlin, Dublin, Ireland

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Case Report A 12 month old boy presented to the Paediatric Emergency Department (PED) with ischaemic necrosis of the distal phalanx of the left thumb as a result of Tourniquet Syndrome.

Six days prior to presentation an adhesive bandage had been applied to the digit to discourage thumb sucking. His mother described crying and irritability, and when the dressing was removed forty-eight hours after application, the infant's mother noted the digit to be shrivelled and mottled. The following day the area began to blister and he was brought to his General Practitioner for review, where hospital attendance was advised.

Examination of the left thumb revealed dry necrosis of the nailbed and distal phalanx extending 1 cm proximal to the nailfold, with an obvious tourniquet mark present at the

interphalangeal joint. The infant was admitted under the joint care of the Plastic Surgery and General Paediatric Teams. The injury was initially managed conservatively. The distal phalanx subsequently auto-amputated and formal surgical debridement and terminalisation at the level of the interphalangeal joint was undertaken. At follow-up twelve months later the injury had healed well with a satisfactory functional outcome.

The severity of the injury coupled with the delay in presentation raised suspicion of neglect or non-accidental injury, and the case was referred to the Medical Social Work Department. The history given by both parents was consistent with the presenting injury. No other concerning injuries or previous hospital presentations were noted and appropriate child-parent interaction was noted throughout. A child protection case conference found that the injury was unintentional and that the infant was not at ongoing risk of significant harm through neglect. A family support plan was put in place on discharge to the community.

Discussion Tourniquet Syndrome is a surgical emergency as circumferential constriction impedes lymphatic drainage with resultant venous outflow obstruction and oedema. Raised interstitial pressures then restrict arterial supply, causing ischaemia and associated pain. If it is not possible to remove the tourniquet in the PED, general anaesthetic and removal in theatre may be required. In cases of significant vascular compromise necrosis and auto amputation may occur as illustrated.

Due to its insidious nature, a high index of suspicion for this condition is required for timely detection. Added vigilance is required in the case of preverbal children and those with speech or learning difficulties who may not be able to express discomfort easily.

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

Rani Wasala, Noman Khan*. Wexford General Hospital, Wexford, Ireland

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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 outpatients 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 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 hypogammaglobulinaemia, alopecia and onychodystrophy. 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 thrombocytopenic purpura. Therefore the patient we reported also has been directed to rule out these endocrinopathies especially adrenocortical deficiencies and auto immune disorders.

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

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

¹Ranya Abuhaleeqa*, ¹Susan O'Connell, ¹Rania Mehanna, ¹John Russell, ²Colm O'Donnell.
¹Our Lady's Children Hospital Crumlin, Dublin, Ireland; ²National Maternity Hospital, Dublin, Ireland

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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 totipotential 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 heterogenous 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 endotracheal 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 thymectomy were done on day 2 after birth. The thyroid gland was not identified surgically. A single parathyroid gland was identified intraoperatively and preserved. The pathology report of the neck mass confirmed immature teratoma arising within and largely effacing the thyroid gland. No malignant germ cell tumour elements identified. He required

levothyroxine treatment to replace the thyroid function. Hypocalcaemia developed postsurgical likely as a result of hypoparathyroidism. He was therefore managed by one alpha and calcium supplements to maintain normocalcaemia. He is feeding orally and continues to show appropriate normal growth and development.

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 hypocalcaemia secondary to hypoparathyroidism developing post operatively should be monitored and treated accordingly.

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P110 THE BOY WITH MOYAMOYA: STROKES AND A WISP OF SMOKE

C Dyson*, K Flinn, E O' Mahony, AM Murphy. Department of Paediatrics, University Hospital Limerick, Limerick, Ireland

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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.