child is under multidisciplinary team involving Paediatrics, ENT, Maxillofacial, Genetics and Speech and Language Therapy team.

Discussion VWS is dominantly inherited with prevalence of 1 in 100,000–200,000. However 30 to 50% of cases arise as denovo mutations hence the diagnosis is made on a clinical basis. Congenital lip pits, cleft lip or palate with varying severity are common presentations in 70% of cases. Submucous cleft palate is a common feature, nevertheless is easily missed on physical examination. Hyper-nasal voice and bifid uvula are isolated in VWS. Hypodontia is a cardinal feature of VWS. Extra-oral manifestations are limb anomalies, popliteal webs and brain abnormalities. Accessory nipples, heart defects, and Hirschsprung’s have also been reported.

Conclusion Overall affected parents carry a 50% risk for each child, and more affected phenotypes gives rise to extreme effects in offspring. However, given more mild features are common, it is prudent to always be vigilant for cleft or lip abnormalities in all offspring. These children need multidisciplinary team approach and long-term follow up.

GP44 ERYTHROPOIETIN AS A TREATMENT MODALITY IN HYPERHAEMOLYSIS COMPLICATING SICKLE CELL ANAEMIA

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Introduction Hyperhaemolysis Syndrome (HS), a severe haemolytic transfusion reaction, is a rare complication in children with Sickle Cells anaemia (SCA) who require transfusion. Donor and autologous red cells are obliterated leading to a worsening of anaemia after transfusion. Erythropoietin has been reported as a treatment modality. This case series examines the experience in our tertiary Paediatric Haematology centre, of treating this haemolytic anaemia with Erythropoietin.

Design and Methods Patients were identified from the SCA patient database at Our Lady’s Children’s Hospital, Crumlin, Dublin in 2018.

Patient charts were reviewed Details of transfusions, presenting symptoms, examination findings, lab results and treatment modalities were recorded.

Results Three children being treated with erythropoietin following HS were identified.

Patient 1: Diagnosed with SCA at birth. Transfusion programme started aged 3 years 6 months due to silent infarct on MRI brain. Transfusion programme stopped aged 3 years 10 months as hyperhaemolysis suspected. Haemoglobin (Hob) fell post transfusion and HbA rose to 20% not 60–70% as would be expected. Autoantibodies were detected.

This was soon complicated by Acute Splenic sequestration with associated high fever. Hob at it’s nadir 5.7 g/L. Erythropoietin commenced with good effect to date.

Patient 2: SCA diagnosed aged 2 years. First transfusion aged 15 years due to crisis with Hob 6. 5 g/L. 5 weeks later admitted with abdominal pain and splenomegaly. Hob 7.45 g/L and then fell to 5.7 g/L. Treated with Immunoglobulin (IVIG) as diagnosis of HS was suspected. Hob A-13.1%. Commenced on erythropoietin and IV Iron.

2 autoantibodies detected. Hob maintained > 6 g/L since then with Erythropoietin 3 times a week and oral iron replacement.

Patient 3: SCA diagnosed aged 1 year. At age 4 years, 3 admissions with chest crises and associated anaemia. Hob fell to a nadir of 4.9 g/L post transfusion and episodes were treated with IVIG and IV methylprednisolone. Another chest crisis at 4 years 7 months precipitated trial of erythropoietin for 3 months with good effect. No further transfusions required. Erythropoietin restarted aged 6 years 6 months due to Hob of 5.7 g/L in association with infection. Hob improved and patient’s Hob now maintained >6 g/L on erythropoietin three times a week.

Conclusion HS is a rare complication of SCA which can cause significant worsening of anaemia that is difficult to treat. Erythropoietin can be used to maintain acceptable levels of haemoglobin to avoid transfusion.

GP45 AUTOIMMUNE ENCEPHALITIS TRIGGERED BY HERPES SIMPLEX VIRUS 1 INFECTION

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Introduction Herpetic encephalitis is the most frequent central nervous system infection in children caused by viral etiology. It may be a trigger for anti-NMDA receptor (NMDAR) encephalitis with the onset of symptoms usually a few weeks after initial presentation with viral encephalitis.

Case report Approximately one third of the patients with herpetic encephalitis develop anti-NMDAR encephalitis. There are more hypotheses on how anti-NMDAR encephalitis may develop. First, herps simplex virus 1 (HSV-1) infection causes an inflammatory destruction of neural tissue with subsequent release of neuronal antigens and formation of NMDAR antibodies; or, second, by molecular mimicry. Onset of symptoms is usually after 2–6 weeks after initial infection with HSV 1. In children symptoms are usually represented by choreoathetosis and orofacial dyskinesias. The diagnosis is made by detecting NMDAR antibodies in cerebrospinal fluid (CSF).

We present the case of an 11 months male infant with no previous medical history, who presented with fever, anorexia, seizures with a focal onset and secondarily generalized, somnolence and progressive neurologic deterioration with onset of symptoms 6 days prior to admission. CSF studies showed a hypertensive, hemorrhagic fluid with low levels of glucose. HSV-1 was detected by real time PCR in CSF. Brain MRI revealed multiple cerebral lesions of different sizes predominantly affecting the right hemisphere, thalamus and basal ganglia with diffuse edema. EEG suggested diffuse cerebral dysfunction. Treatment with Acyclovir was started and continued for 28 days with notable decrease in
symptom severity. Fever reappeared approximately at 2 weeks after admission. He presented neurologic deterioration with focal seizures, choreiform movements, facial dyskinesia, psychomotor regression to the age of 2 months. These symptoms were suggestive of an autoimmune encephalitis. NMDAR antibodies were detected in CSF. Recurrent HSV encephalitis was excluded. Steroids and Intravenous Immunoglobulins were associated to the therapy scheme. The patient recovered slowly and partially but remained with neurological sequelae.

Discussion Our case exemplifies an anti NMDAR encephalitis which developed in the convalescence phase of a viral infection. HSV-1 infection may be a trigger for anti-NMDAR encephalitis. Autoimmune encephalitis after viral infection is important to be recognized early as patients can receive treatment with steroids and immunosuppressants in order to improve their outcome.

**GP46**

CONGENITAL HYPERINSULINISM: IMPORTANCE OF EARLY DIAGNOSIS BY GENETICS & F-DOPA SCANNING FOR OPTIMAL MANAGEMENT: 2 SEPARATE CASE REPORTS

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Background Congenital hyperinsulinism (CHI) is a rare inherited disease (1 in 50,000 newborns), that can be diffuse or focal. Mutations in ABCC8 gene are the most common cause. Advances in molecular genetics, imaging techniques (18FDOPA-PET-CT) and surgery have radically improved the outcome

Objective To report two cases of infants with CHI and highlight how early genetic results aided medical and surgical management

Case 1 A 5.68 kg, baby boy born at term via normal delivery had hypoglycaemia (blood glucose (BG) 0.8 mmol/L) at 2 hours of life. He required a glucose load of 20.2 mg/kg/min, 25% dextrose and fluid volume 200ml/kg/day. At BG <2.6 mmol/L Insulin was 180 pmol/L (< 14 pmol/L) in the absence of ketosis. He was unresponsive to maximum dose of Chlorothiazide & Diazoxide (20 mg/kg/day) and Octreotide 10mcg/kg/day. He had difficult IV access and required a PICC line. Subsequently he was commenced on glucagon infusion prior to transfer to The Northern Congenital Hyperinsulinism Service (NORCHI), U.K.

Case 2 A 4 kg, term, male baby, forceps delivery was admitted to NICU on day 1, with hypoglycaemia (blood glucose (BG) of 1.4 mmol/L). Despite increasing glucose load of > 18 mg/kg/min, Dextrose 20% and fluid volume 180ml/kg/day, he continued to be hypoglycaemic. At BG of 0.9 mmol/L Insulin was 170 pmol/L (< 14 pmol/L) without ketosis. He was commenced on Diazoxide & Chlorothiazide but considered unresponsive, due to ongoing hypoglycaemia. Furthermore, he received a glucagon infusion 12.5mcg/kg/hr & transferred to NORCHI where his Diazoxide was stopped while Glucagon & Octreotide continued. In both cases, early genetic samples were sent from CUH. Thus, by the time they reached NORCHI, genetic results were available: both cases were heterozygous paternally inherited mutation in ABCC8 gene. F-DOPA scanning revealed a focal lesion in the body and head of pancreas for the first and second case respectively. A laparoscopic focal pancreatic lesionectomy was performed in both cases. The post-operative period for the first case was uneventful with no hypoglycaemia, discontinuation of Glucagon & Octreotide providing complete cure without medications. Post-surgery the second case required subcutaneous Octreotide 10mcg/kg/day in four divided doses, due to residual hyperinsulinemic tissue. At three years of age repeat F-DOPA scan revealed no residual lesion. Subsequently, he was weaned off Octreotide without any further hypoglycaemia.

Conclusion In both cases early detection of CHI with early genetic testing, F-DOPA scanning and collaboration with centres of excellence like NORCHI, all optimised the care & appropriate management.

**GP47**

A NOVEL MUTATION IN AUTOSOMAL DOMINANT SPASTIC PARAPLEGIA TYPE 4

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Background Hereditary Spastic Paraplegia (HSP) is a rare group of diseases characterized by slowly progressive degeneration of the corticospinal tracts. It can present at any age with varying degrees of lower limb spasticity and weakness. HSP syndromes are classified as “complicated” when associated with other neurologic features such as muscle atrophy, ataxia or cognitive impairment. Mode of inheritance can be autosomal dominant, recessive or×linked and more than 80 genetic types of HSP have been identified. Usually diagnosis is delayed due to its low prevalence and variable clinical phenotypes.

Method A 3 year old female child, born to non-consanguineous parents presented at 17 months with delayed walking. She was walking with both hands held and her gait pattern looked abnormal. She had had a previous normal hip ultrasound examination at 6 weeks of age as there was a history of breech presentation. Hip X-rays later ruled out developmental dysplasia of hips. She attended physiotherapy and at two years of age was not still walking independently. She was a term infant with unremarkable perinatal history. Her father and a paternal cousin had a diagnosis of ‘cerebral palsy’. An older male sibling is asymptomatic. Examination revealed weight bearing on her forefeet with lower limbs turning to internal rotation suggestive of a diplegic gait. There was mildly increased tone in her lower limbs with bilateral brisk reflexes and extensor plantars. Upper limb examination was normal. Developmental examination was age appropriate. While she had normal hip x-rays and MRI scan of the brain, the family history prompted further investigations looking for a familial cause for her clinical picture.

Results Genetic panel for HSP identified a novel heterozygous mutation in exon 12 of the SPAST c.1478A>T, p(Asp493Val) gene consistent with a diagnosis of autosomal dominant spastic paraplegia type 4. This is considered as an ‘uncomplicated’ HSP where symptoms might not progress over many years. The same genetic mutation was confirmed in her father too.

Conclusion The clinical findings and family history led to targeted genetic testing, subsequent early diagnosis and appropriate genetic counselling of family members.