sclerosis, optic neuritis, transverse myelitis or NMO spectrum disorders[1,2]. Associated morbidity and mortality of each is vastly different. Gaining accurate information through appropriate investigation is vital for appropriate treatment and counselling. There is currently no national or international guideline for investigation of an acute demyelinating episode.

Methods Retrospective review of all cases of a first demyelinating event over a 10 year period (2008 – 2018) at Cork University Hospital. Laboratory investigations, imaging and clinic letters were reviewed.

Results In total eighteen cases were reviewed. Eventual diagnoses were 7 ADEM, 4 ADEM with transverse myelitis, 4 Multiple Sclerosis and 3 Optic Neuritis. Median presentation age was 6 years (1 year 4 months - 15 years 10 months). WCC and CSF microscopy was done in 100%. CRP and ESR done in 89% and 28% respectively. Investigations for bacterial and viral causes either on serum, CSF or swabs was inconsistent varying between 11–83%. CSF antibodies, including anti MOG, anti-NMDA, Aquaporin 4 and anti voltage gated potassium channel antibodies were sent in 6–39% of cases dependant on test. Oligoclonal bands were sent in 83%. Imaging was undertaken in all cases with seventeen having an MRI Brain. Median time to MR brain was 1 day (0 days – 6 days). Fourteen cases had a MR spine with median time to spinal imaging of 2 days (1 day – 11 days).

Conclusion This review highlights the variable approach to investigation of suspected demyelination. The wide differential and need for prompt treatment to prevent long term neurological disability means there are multiple complex investigations required within a short time period. The laboratory investigations and neuroimaging required are labour intensive and incur significant financial cost. This is of particular importance in children, many of whom will require sedation and at times general anaesthetic to ensure successful obtaining of samples. The availability of a local protocol would guide clinicians investigation when faced with an unfamiliar presentation under significant time pressure. It would ensure appropriate and timely investigation enabling appropriate treatment and counselling.

GP232 MANAGEMENT OF SEIZURES IN CHILDREN WITH THERAPY-RESISTANT EPILEPSY

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Epilepsies in children represent a heterogeneous group of disorders and syndromes with different etiology, severity, prognosis and treatment. Early diagnosis, accurate recognition of underlying aetiologies leads to more effective management and treatment and improve overall health and quality of life. Genetic testing is very important in the cases of therapy-resistant seizures. The purpose of study was to recognise the possible reason of failed AED treatment and to find the ways to owercome it.

Methods 45 patients with different forms of epilepsy aged from 3 months to 16 years not the candidate for surgical treatment have been studied. The long duration EEG, high resolution MRI, blood biochemical tests, blood level lactate and ammonia, amino acid, organic acid and disturbance of fatty oxidation by TMS, genetic investigations (mtDNA and exome sequence), measurement of autoantibodies to NR2 and GluR1 in blood serum by ELISA were performed to these children.

Results The respiratory chain disorders confirmed by mtDNA sequence were found in 11 children. Metabolic epilepsies discovered in patients have the following origins: two with glutaric aciduria type1, one – glutaric aciduria type2, one with propionic aciduria, one with methylmalonic aciduria, one with Gaucher's disease type3, two patient with glycogenosis type 9, two patients with ceroid lipofuscinosis type 2 and 6,lysosomal storage disorders in 3 cases.

Genetic epilepsies with mutation in genes SCN8A (two patients), GRIN2A, KCNMA1, SRPX2, SCN9A, ACO2, ARHGEF9, 15q11.2q13.3,TSC- 4 patients were revealed. In other cases with normal MRI the reason of pharmacoresistant seizures was not discover yet. The elevated level of autoantibodies to glutamate NR2 and GluR1 receptors were found in children of these groups. But in patients with metabolic epilepsies the elevation level of autoantibodies to NR2 was in 4 to 7 times higher in comparing with children with genetic epilepsies. In children with metabolic disorders and energy metabolism disorders we use the specific therapy (special diet, L-carnitine, vitamins, enzyme replacement therapy etc) in cases which it possible, avoid valproic acid in treatment of children with mitochondrial disorders and glutaric acidurias, as well we use the phenytoin in patient with potassium channel mutation SCN8A. These treatment management led to reduction in seizures frequency or even to seizures remission in some cases.

Conclusions The recognition and diagnostic of underlying etiologies of intractable seizures improve the treatment management in many cases. The excessive NMDA transmission might be the part of pathogenesis of seizures in children with inborn error of metabolism.

GP233 LIFESAVING MECHANICAL THROMBECTOMY IN PAEDIATRIC STROKE

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Introduction Childhood arterial ischaemic stroke (AIS) is uncommon with a reported incidence between 1.2 and 7.91 per 100,000 per year.[1,2,3] Previously it was thought that children with AIS had a good outcome due to brain plasticity; however, mortality has been reported in up to 28%, and morbidity in up to 70% of survivors.[4,5] There are no randomised trials of mechanical thrombectomy in children. The 2017 published RCPCH stroke guidelines draw on the excellent outcomes for mechanical thrombectomy in adult trials and recommend referral for intra-arterial clot extraction in patients with NIHSS score of 6 or more and up to 12 hours post onset if there is salvageable brain tissue on imaging.[6] There are only 29 paediatric cases published in the literature that have undergone mechanical thrombectomy, 12 of which were for posterior circulation AIS.

We describe a case of a 3-year-old girl with bilateral pontine and cerebellar infarction due to basilar artery thrombosis, related to a diagnosis of exclusion of severe iron deficiency anaemia who was successfully treated by mechanical thrombectomy.

Case Report Our patient presented with a one-day history of vomiting, and headache, on a background history of a viral prodrome the preceding week, and varicella infection three months earlier. Her haemoglobin was 4.6 g/dL, with a profoundly microcytic, hypochromic blood film. She received a blood transfusion with clinical improvement. At 36 hours after admission, she became irritable and developed a left divergent squint (NIHSS score 4). Neuroimaging demonstrated acute infarction of the pons, cerebellum and punctate lesions in both occipital lobes. MRA showed complete occlusion of right vertebral artery and basilar artery. Enoxaparin was commenced, but 12 hours later she developed left CNVII palsy and a dense left hemiparesis with hypertension. CT Brain showed worsening ischaemia. Given her ongoing clinical deterioration with major risk of significant morbidity and indeed mortality she underwent mechanical thrombectomy. Successful recannalisation of the basilar artery occurred with a distal left parieto-occipital thrombus remaining. At 9 months follow-up she has a mild left hemiparesis, left CN VII, III and IV palsies and right CNVI palsy, but mobilises independently and is on a normal diet (mRs 2); imaging shows established pontine infarction with gliosis.

Conclusion This case adds to the limited reports of mechanical thrombectomy in children in posterior circulation AIS, as a safe and effective treatment. It also highlights the importance of recognising severe iron-deficiency anaemia as a cause for AIS in children.

GP234 THROMBOLYSIS OF ARTERIAL ISCHAEMIC STROKE IN AN EIGHT MONTH OLD: THE FIRST IRISH CASE

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Paediatric arterial ischaemic stroke (AIS) is a potentially fatal condition with serious long term neurological sequelae. The rate of paediatric ischaemic stroke is 1.6 – 3.3 per 100,000 and 61% of these children are left with long term neurological difficulties. Thrombolysis guidelines for children were introduced in May 2017 by the Royal College of Paediatrics and Child Health (RCPCH). However, these guidelines were developed for children from the age of eight and up, and may be used on a case by case basis for children from the age of two to eight years. There is very limited data on the use of thrombolysis in children of any age, particularly in the under two age group. Here we present the first case of AIS thrombolysis in an under two year old in Ireland.

An eight-month old boy presented with a two-and-a-half-hour history of decreased movement of his left side. Importantly, his mother had noted the exact time symptoms commenced. His brain imaging showed a large right sided area of ischaemia in the territory of the middle cerebral artery (MCA), and a narrowed right internal carotid artery (ICA); no haemorrhage was seen. He was brought to intensive care and thrombolysis was commenced with alteplase and heparin. Methylprednisolone was also given as the narrowing of the ICA was felt to be inflammatory. Other investigations, including echocardiogram, homocysteine level, thrombophilia screen and varicella titres, were all negative.

He has recovered well. He is back on full oral feeds and has learned to walk. He is right hand dominant, with no left sided neglect. He has no speech delay and has been discharged by both physiotherapy and occupational therapy.

Enrolling children in randomised controlled trials to assess AIS treatment and outcomes remains extremely difficult, as evidenced by the Thrombolysis in Pediatric Stroke (TIPS) study, which was closed by the National Institute for Health (NIH) after only thrombolysing one patient despite recruiting 43. The issues centre on delays in presentation (usually outside the thrombolysis window), and in delayed recognition of AIS in emergency departments by clinicians. This case highlights the need for continued research in this area as it demonstrates that successful outcomes are possible after thrombolysis of AIS.

GP235 COL4A1 MUTATION INHERITED FROM MATERNAL MOSAICISM IN AN INFANT PRESENTING WITH MICROCEPHALY, HAEMOLYTIC ANAEMIA AND CATARACTS

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Introduction *COL4A1*-related disorders have overlapping signs and symptoms as a result of fragile blood vessels. The COL4A1 gene located on Chromosome 13 is responsible for making the alpha one subunit of type IV collagen. Type IV collagen molecules form complex protein networks, which are the main component of basement membranes surrounding the body's blood vessels. Clinical presentation and the onset of symptoms are widely varied and largely dependent on the phenotype expressed. A number of phenotypes with overlapping features have been described. These include COL4A1 Brain Small Vessel Disease, AD Familial Porencephaly and HANAC(Hereditary Angiopathy with Nepropathy, Aneurysms and Muscle cramps) Syndrome. COL4A1 disorders are inherited in an autosomal dominant manner and the proportion of cases caused by *de novo* pathogenic variant is estimated to be 27%.

Case presentation A female infant was born at 36+2 weeks gestation via Emergency LCSC for IUGR and reduced fetal movements. Apgars were normal. BW was 1.88 kg(2nd centile) and Birth OFC 29.7cm(3rd centile). Initial exam was normal and the infant was admitted to the SCBU for Low Birth weight. She developed haemolytic anaemia week two of life and required three RCC transfusions on DOL 18, 26 and 35. Head growth was found to be static on week four of life and she was also noted to have absent red reflexes bilaterally. Cranial Ultrasound showed a large cystic abnormality in the right cerebral hemisphere. MRI Brain showed cystic encephalomalacia with a large right hemispheric cyst, simplified gyral pattern, significant volume loss and evidence of previous haemorrhage in left cerebral hemisphere. Opthamology confirmed bilateral cataracts. Extensive investigations including metabolic screen, infectious workup, Karyotype and and microarray were all normal. Further genetic testing revealed that the child was heterozygous for novel COL4A1 missence