Childhood arterial ischaemic stroke is rare, with an incidence of 1.6 per 100,000.¹ As demonstrated in our patient, early recognition is key for thrombolysis to be effective. The publication of the Stroke in Childhood: Clinical guideline for diagnosis, management and rehabilitation (2017)² advocates the importance FAST (Face, Arms, Legs and Time) which has already proved to be successful for early diagnosis in adult patients. Effective collaboration with our adult counterparts is vital to share resources and implement guidance locally.

REFERENCES

1. Mallick AA, Ganesan V, Kirkham FJ, *et al.* Childhood arterial ischaemic stroke incidence, presenting features, and risk factors: A prospective population-based study. *The Lancet Neurology* 2014;**13**(1):35–43.
2. Stroke in childhood: Clinical guideline for diagnosis, management and rehabilitation. *Royal College of Paediatrics and Child Health* 2017.

G314(P) A RARE NEUROLOGICAL CAUSE OF LIMPING AND HIP DYSPLASIA

A Butt, G Sinha. *Paediatric, Walsall Manor Hospital, Walsall, UK*

10.1136/archdischild-2018-rcpch.305

Introduction Limping is a common presentation in children. It has many aetiologies, from trauma, infection, haematological conditions and malignancies. We would like to present a rare cause of limping. Syringomyelia describes a longitudinal fluid cavity of the spinal cord, whether a dilatation of the central canal or cavitation of the grey matter. There is a paucity of data, particularly of paediatric cases. Clinical manifestations depend on the location and size of the syrinx. They typically involve segmental signs, with wasting of the small muscles of the hand, sensory deficits and absence of tendon reflexes. It can also cause lower limb spasticity, and a loss of pain and temperature sensation.

Case report A previously fit and well 18 month girl was referred with a 3 month history of dragging the left foot and limping. The mother's pregnancy and child's neonatal period was uneventful. Her growth was between the 9th-25th centile, and developmental milestones were appropriate. She was passing urine normally, and was constipated for 4 months. On initial assessment she was noted to have birth marks on her lumbar spine. She had normal tone, power and reflexes in both lower and upper limbs. She had no wasting of the small muscles of the hands. Her left foot appeared to be larger than the right, and when walking she was dragging her left foot. At this point the parents were advised how to manage the constipation and the child was sent for physiotherapy, hip X-rays and MRI of the brain and spine. The hip X-ray showed mild loss of coverage of the left femoral head. The cranial MRI was normal. MRI without contrast showed a cystic abnormality in the conus medullaris most likely due to Syringomyelia; MRI contrast excluded spinal tumour. The radiologist felt the abnormal development of the left hip could be secondary to the syrinx, or evolution of developmental dysplasia.

Learning points/conclusion This case highlights the need for full examination, and appropriate investigation of the spine, hips, abdomen and lower limb from a musculoskeletal and neurological perspective.

G315(P) TREATMENT OF ANTICARDIOLIPIN ANTIBODY MEDIATED CHOREA IN AN ADOLESCENT MALE

¹R Odedun, ¹R Kumar, ²L McCann. ¹Paediatric Neurology, Alder Hey Children's Hospital, Liverpool, UK; ²Paediatric Rheumatology, Alder Hey Children's Hospital, Liverpool, UK

10.1136/archdischild-2018-rcpch.306

Background Anti-phospholipid antibody associated chorea is a rare cause of this movement disorder, with a higher incidence in females and children. Other causes include Wilson's disease, Sydenham's chorea and ataxia-telangiectasia. Aetiology is uncertain, but may involve anti-phospholipid mediated dysfunction of the basal ganglia. We report an unusual case of anticardiolipin antibody related chorea in an adolescent male presenting to secondary care services.

Case report Our patient is a 14 year old male who became unwell in December 2016 with gradual onset of uncontrolled movements in his arms, and behavioural changes. There was a notable history of minor head injury, preceding sore throat and recurrent tonsillitis. Over the next six months, he reported worsening of choreoathetoid movements affecting his gait and upper limb function.

He presented in April 2017 with acute slurring of speech three weeks after starting baclofen. He was then referred to tertiary paediatric neurology services. No features of Lupus were present on examination. His functioning was monitored after baclofen was stopped, and he progressed to treatment with high dose steroids, penicillin V and sodium valproate. He improved following significant rehabilitation with multidisciplinary involvement from occupational therapy, physiotherapy and the school team. His final diagnosis was revised to anticardiolipin antibody related chorea following results of immunological tests and consideration of the chronic progressive history. He was treated successfully with aspirin and mycophenolate mofetil, and has been discharged home with ongoing clinic review.

Results 12 lead ECG, echocardiogram and MRI brain normal. ASO Titre: 400 units/ml (50–200)
Anti-DNAse B: 100 units/ml (<240 units/ml)
Anti-Mitochondrial Ab: positive
Anti-Nuclear Ab titre: 1:320 (Positive)
Anti-cardiolipin IgG Ab 1184.2 units/ml (0–19.9)
Anti-B2-GP-1 IgG 4269.7 units/ml (<20)

Conclusion As a patient's disease continues to evolve, so should our diagnostic approach. Atypical progression of disease should prompt review. Generally, treatment of anticardiolipin related chorea falls into 2 pathways; anticoagulation and immunosuppression, with evidence limited to case reports and small case series.

Acknowledgements to the local medical illustration team for performing serial videos showing improvement in function.

G316(P) COPY NUMBER VARIATION (CNV) IN A PATIENT WITH EPILEPSY AND HYPOTHYROIDISM: A RARE ASSOCIATION WITH RBFOX1 MICRODELETION

S Kapoor, S Kumar, N Hussain, S Ganesan. *Department of Paediatrics, University Hospitals of Leicester, Leicester*

10.1136/archdischild-2018-rcpch.307

Introduction The effect of this deletion has an impact on development of various organs and glands. 16 p13.3 microdeletion is a rare genetic condition with a variable phenotype spectrum. *RBFOX1* [RNA binding protein] located on chromosome 16 p13.3, is one of the three members in Fox gene family encoding splicing factors. We would like to present this case report to elucidate the association of this alteration with hypothyroidism and epileptic seizures as only one case of this association has been reported in the literature.

Case report An 8 year old boy presented with recurrent afebrile, unprovoked seizures and behaviour difficulties. EEG was supportive for multifocal epilepsy. The MRI brain scan was normal. Blood results showed persistently high TSH levels and low free T4. Subsequent workup showed a high TPO level and the endocrine team made a diagnosis of autoimmune hypothyroidism. Genetics investigations showed that the patient had a 16 p13.3 microdeletion. A similar genotype was noted in the mother though she was phenotypically normal.

Discussion *RBFOX1* is an important splicing factor identified as a binding protein of *ATXN2*, suggesting a role in neurologic function. It is highly expressed in the brain, skeletal muscle, heart, other organs and glands. Any alteration with this specific locus is associated with variable phenotypes affecting various body systems. These include mental retardation, epilepsy, mental health disorders, congenital heart defects, obesity and diabetes along with other endocrine problems. The introduction of array comparative genomic hybridization (CGH) has provided the ability to map DNA copy number variations (CNVs) genome with high resolution. Only one patient with this CNV has been reported to have congenital hypothyroidism and associated epilepsy. In our patient though the CGH array has helped in mapping out the genetic defect however its role in hypothyroidism is yet to be established.

Conclusion Patients presenting with multiple system involvement, especially involving the endocrine system with associated developmental delay and neurobehavioral and neuropsychiatric problems should undergo CGH array analysis as part of their endocrine assessment. Moreover endocrinologists need to be aware of this copy number variation as it may have a huge impact on future pregnancies of the patient.

G317(P) ABSTRACT WITHDRAWN

G318(P) SCN1A-RELATED EPILEPSY: THREE DIFFERENT CASES AND A LITERATURE OVERVIEW

¹T Ratnaik, ¹K Moodley, ¹A Saraswatula, ²G Ambegaonkar. ¹Paediatric Department, West Suffolk Hospital, Bury St Edmunds, UK; ²Department of Neurology, Addenbrookes Hospital, Cambridge, UK

10.1136/archdischild-2018-rcpch.308

Among epilepsies caused by a single gene mutation, the sodium channel neuronal type 1 α subunit (SCN1A) gene, encoding the voltage-gated sodium channel Na_v 1.1, is the most frequently associated with epilepsy (Brunklaus *et al.* 2014).

The mutations cause a spectrum of disease including genetic epilepsy with febrile seizures plus (GEFS+), Dravet syndrome or severe myoclonic epilepsy of infancy (SMEI), and borderline severe myoclonic epilepsy of infancy (SMEIB). We describe three children diagnosed with SCN1A-associated atypical epilepsy phenotypes whose treatment continues to be challenging. We report 3 children less than 12 months presenting with multiple pharmacoresistant seizure types proving diagnostic and clinical challenge. All present with a heterozygous sequence change identified in SCN1A gene.

As per McDonald and colleagues (2017), carbamazepine, oxcarbazepine, and lamotrigine may worsen focal seizures associated with SCN1A mutation, or have other adverse effects. Sodium valproate, stiripentol, and clobazam are thought to have the best impact on management of focal seizures such as those seen in Dravet syndrome. Carbamazepine was also seen to exacerbate seizures in GEFS +epilepsy positive for SCN1A mutation (Shi *et al.* 2016).

The establishment of the SCN1A variant database may aid further therapy development targeted to specific genetic mutations (Claes *et al.* 2009). Cannabidiol, ketogenic diet, fenfluramine therapies are novel therapies used in the treatment of this highly pharmacoresistant form of epilepsy.

SCN1A-related epilepsy consists of a spectrum of disease that is still difficult to manage due to the different seizure types present and treatment resistance. This is certainly true of our three cases, all three are female children who presented in infancy and have suffered from an evolving clinical course. They are all being treated with stiripentol, however, have not achieved seizure control, and it will be vital to keep updated with ongoing research into newer therapies.

G319(P) OUTCOMES FOLLOWING FEBRILE CONVULSIONS: A RETROSPECTIVE COHORT STUDY

¹L Chatterton Dickson, ^{1,2}J Shetty. ¹Edinburgh Medical School, University of Edinburgh, Edinburgh, UK; ²Child Life and Health, Royal Hospital for Sick Children, Edinburgh, UK

10.1136/archdischild-2018-rcpch.309

Febrile convulsions (FC) are seizures in children under five associated with fever. They are reported in 2%–5% of children with good prognosis. However, the risk of epilepsy may be increased. This study reported the first Scottish cohort and observed current clinical practice, outcome and associations with socioeconomic status. A FC database was created using routinely collected clinical data of all children attending A and E. A Scottish prevalence of 390/100,000 children/year was elucidated with 2.6% of children experiencing FCs by their fifth birthday. Children from deprived areas were overrepresented. It is established that only very young children or children with complex convulsions should be admitted and further investigations are usually unnecessary. However, 72.6% of children were admitted and 10.4% received an EEG which is inappropriately high. Epilepsy was diagnosed in 2.6% of children, an increase from the 1% population risk. However, many of these children had other abnormalities. Only 1.5% of otherwise healthy children developed epilepsy. Overall, the danger of FCs should not be overstated as they only slightly increase the risk of epilepsy in an otherwise healthy child. This should be explained to quell parental anxieties and reduce unnecessary admissions and investigations.