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G317(P) ABSTRACT WITHDRAWN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

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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

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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.

G320(P) POST-MALARIA NEUROLOGICAL SYNDROME: THE FIRST IRISH PAEDIATRIC CASE

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10.1136/archdischild-2018-rcpch.310

Aims Post-malaria neurological syndrome (PMNS) is described as a rare post-infectious encephalopathy occurring within two months of resolved malaria infection and with an aparasitaemia. PMNS encompasses three separate neurological syndromes: A delayed cerebellar syndrome, an acute demyelinating polyneuropathy (GBS) and an acute disseminated encephalopathy (ADEMs).

Here, we report the first Irish paediatric case of falciparum PMNS, in a patient of African origin, born and living in Ireland.

A 15 year old boy presented with a 3 day history of progressive encephalopathy, features of raised ICP and seizures on a background of falciparum malaria treated six weeks previously. PMNS was diagnosed after further investigations and an aparasitaemia. He was sedated and intubated for 2 days and commenced on antimicrobials, antimalarial and steroids. His investigations results as following: MRI brain: Cerebral oedema and optic neuritis, EEG: Severe encephalopathy. Serial thick and thin films: No malaria parasites Falciparum Protein Antigen (RDT): Positive (Can remain positive for 6 weeks after malaria). CSF Studies: Protein 1383 g/dl, 16 WBCs/dl, 100% mononuclear cells). PCRs: negative for HSV, Adenovirus, Coxsackie, EBV, CMV, Meningococcus, Pneumococcus.

By day 6 of admission he had made a full recovery with no neurological deficits.

Methods Using Google and PubMed, we searched for relevant case reports and journal articles describing neurological syndromes occurring post infection with falciparum malaria in the paediatric population.

Results Whilst the prevalence of PMNS (plasmodium falciparum) is 0.12% in adults, the prevalence in children remains unknown. In 1996, a Vietnamese study conducted over 4 years reported 23 patients with PMNS following full recovery from falciparum malaria. Of these, only 3 were children. A 2015 case report describes a further two children with falciparum PMNS. There have been no further paediatric cases reported to date worldwide.

Conclusion In conclusion, PMNS is an increasingly recognised, but rare complication of malaria that must be differentiated from relapsing or recurrent malaria, and post-infectious neurological syndromes, e.g. ADEM. In particularly severe cases, steroids have been given as an adjunctive therapy to speed recovery however PMNS is a self-limiting condition that resolves within 2–14 days and requires no specific treatment.

G321(P) SPONTANEOUS REMISSION OF EPILEPSY IN A CASE OF MESIAL TEMPORAL SCLEROSIS

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10.1136/archdischild-2018-rcpch.311

Background Mesial temporal sclerosis (MTS) is commonly referred to as hippocampal sclerosis (HS), is the most

common association with intractable temporal lobe epilepsy. The typical MRI features of hippocampal sclerosis, are unilateral volume loss and increased signal intensity on T2-weighted images. There is no sex or side preference and a proportion of cases are bilateral. Macroscopically the hippocampus is firm and shrunken. Microscopic findings include a characteristic pattern of neuronal loss and reactive gliosis that varies in severity from case to case. Mesial temporal sclerosis is an uncommon finding in children, but when it occurs, it is always associated with epilepsy. Mesial temporal sclerosis is the most frequent cause of drug-resistant temporal lobe epilepsy but has a satisfactory response to surgery, and is considered infrequent in children.

Patient Characteristics: 13 year old girl, she was born at 41 week of gestation with an uneventful antenatal history. She was ventilated at birth and also treated for streptococcal meningitis. She developed seizures during this treatment period. She didn't require antiepileptic at discharge. No developmental concerns. She presented with focal seizures at the age of 3 years. Had further seizures, about one or two a year, up to the age of 6 years. Eye deviation to right side, abnormal mouthing movements, disoriented, confused, vacant, lip smacking lasting for 10 min followed by headache and vomiting with post ictal phase for two hours. She remained seizure free for the past 6 years. She was not commenced on any medication.

Investigations EEG at the age of 5 years shown clustered spike epileptiform discharges focal in the central, temporal and occipital area.

MRI scan – MRI brain scan showed dilatation of the left temporal horn with left mesial temporal sclerosis.

Conclusion This is a case of mesial temporal sclerosis with spontaneous remission of epilepsy. It is extremely unusual for mesial temporal sclerosis to be spontaneously seizure free. Pathophysiological mechanism of epilepsy in mesial temporal sclerosis is not fully understood. Further imaging and pathological studies comparing similar cases against the usual MTS with drug resistant epilepsy might lead to find clinically useful indicators.

G322(P) NOT BELL'S PALSY ANYMORE? LYME DISEASE (LD) UNTIL PROVEN OTHERWISE

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10.1136/archdischild-2018-rcpch.312

Aims Lyme Borreliosis (LD) is becoming increasingly prevalent across parts of the UK. Recent evidence suggests that LD is the commonest cause of lower motor neuron type facial palsy (LMN FP) in children and adults in the USA. Historically, idiopathic LMN FP, termed Bell's palsy, was given as the commonest cause in the UK, we discuss whether current evidence suggests otherwise.

Methods We report 2 cases of LMN FP seen over an evening shift, which were subsequently serologically confirmed cases of LD. We also reviewed current literature and surveillance data.

Results LD is an infectious disease caused by the spirochaete *Borrelia burgdorferi*. It is the most common tick-borne infectious disease in the UK and is becoming increasingly prevalent in certain areas, affecting around 9.8/100 000, a figure that continues to rise. The presenting features are often non-