Patient X is a 3 year old male who had neonatal hypoglycaemia and confirmed homozygosity for the GSD Type IIIa gene. Patient X was commenced on the KD at 8 months of age when progression of his GSD resulted in severe cardiomyopathy. The following outlines the transition onto the KD and clinical findings between 8-22 months

Method Standard dietetic treatment was provided from birth to maintain euglycaemia along with placement of a percutaneous endoscopic gastrostomy. Continuous 24 hour feeding was required due to unsuccessful bolus feeding. A modified KD incorporating MCT fat -which has been noted to further aid ketosis was commenced at 8 months with a ketogenic ratio of 0.5:1 building to 1:1 within 2 weeks. Ketones and blood sugar levels were closely monitored with a threshold of 2.6 mmol/L of glucose and ketones >1 mmol/L before hypoglycaemia intervention was required.

Results Prior to the KD, there was a high glucose infusion rate (GIR) of 9.75 mg glucose/kg/minute. After initiation of the KD, the GIR reduced initially to 5.8 mg glucose/kg/minute, with a gradual increase of MCT fat from 6% to 28% and the GIR further reduced to 2.73 mg glucose/kg/minute. There were no episodes of hypoglycaemia and ketones ranged from 1–2.9 mmol/L. Echocardiographs showed a significant improvement in cardiac function with a cardiac output reduction of 137 to 39 mmHg.

Discussion The KD was trialled as an alternative treatment. It resulted in the reduced intake of carbohydrate and the subsequent reduction of glycogen build-up within cardiac muscle. Ketones were used as an alternative fuel source and euglycaemia was maintained

Conclusion The KD should be considered as an alternative treatment for GSD Type IIIa where standard intervention is not effective.

GP230 FETAL ACETYLCHOLINE RECEPTOR INACTIVATION DUE TO MATERNAL MYASTHENIA GRAVIS: AN UNDERRECOGNISED, DEVASTATING BUT POTENTIALLY PREVENTABLE AND TREATABLE DISORDER

¹Mark O'Rahilly*, ²Andreas Hahn, ³Cam-Tu Nguyen, ⁴Dae-Seong Kim, ⁵Shin Y Byun, ⁶Ulrike Schara, ⁶Maria Henrich, ^{7,8,9}Jacob Leslie, ^{7,8,9}Angela Vincent, ¹Nicholas M Allen*, ^{10,11,12}Heinz Jungbluth*. ¹Department of Paediatrics, Galway University Hospital/National University of Ireland, Galway, Ireland; ²Department of Child Neurology, Feulgenstr, Giessen, Germany; ³Clinical Neurological Sciences, Children's Hospital, London Health Sciences Centre, London, Ontario, Canada; ⁴Department of Neurology, Pusan National University School of Medicine, Pusan, Korea, Republic of; ⁵Department of Paediatrics, Pusan National University School of Medicine, Pusan, Korea, Republic of; ⁶Department of Paediatric Neurology, Developmental Neurology and Social Paediatrics, University of Essen, Essen, Germany; ⁷Department of Clinical Neurology, Oxford University, Oxford, UK; ⁸Department of Clinical Neurology, John Radcliffe Hospital, Oxford, UK; ⁹Department of Clinical Neurosciences, Weatherall, Institute of Molecular Medicine, University of Oxford, Oxford, UK; ¹⁰Department of Paediatric Neurology, Neuromuscular Service, Evelina's Children Hospital, Guy's and St. Thomas' Hospital NHS Foundation Trust, London, UK; ¹¹Randall Division for Cell and Molecular Biophysics, Muscle Signalling Section, London, UK; ¹²Department of Basic and Clinical Neuroscience, IoPPN, King's College, London, UK

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Aim Fetal acetylcholine receptor inactivation syndrome (FARIS) occurs in offspring of mothers affected by myasthenia gravis (MG), from in-utero exposure to acetylcholine receptor (AChR)-antibodies targeting the fetal AChR γ -subunit. FARIS causes damage to the fetal neuromuscular junction which is crucial in muscle development, causing a persistent myopathy. FARIS may initially be mistaken for Transient

Neonatal Myasthenia Gravis (TNMG), congenital neuromuscular disorders and one of the many causes of neonatal hypotonia. This study aimed to determine the clinical spectrum of FARIS and assess oral salbutamol as a novel pharmacological therapy.

Methods Detailed review of antenatal and postnatal clinical features in novel FARIS cases seen in international neuromuscular centres. Antibody data analysis was performed at the Oxford neuroimmunology research laboratory. Oral salbutamol was trialled in five cases based on previously reported benefit in one of our patients.

Results We identified 12 novel FARIS cases. At delivery resuscitation was required in all and intubation in nine, all had severe generalised hypotonia. Two infants with arthrogryposis-multiplex-congenita phenotype died in the neonatal period. Among survivors, there was requirement for mechanical ventilation (n=9), NIPPV (n=2), oxygen (n=1), and supplemental NG/PEG feeding (n=12). The presence of severe generalised hypotonia with dysmorphic features prompted investigations for other neuromuscular, genetic and metabolic disorders which were negative. Common features included facial weakness (n=12) and limb contractures (n=9). Newly described disease features: diaphragmatic paresis (n=5), hearing impairment (n=3), CNS involvement (n=3), pyloric stenosis (n=2), extra-ocular eye restriction (n=2), non-progressive scoliosis (n=2), and jaw opening contracture (n=1). Motor development of patients improved with time. Respiratory complications (tracheostomy; n=2), feeding difficulties (PEG; n=2), facial weakness and speech impairment (from velopharyngeal incompetence) persisted in most. TNMG treatments (immunotherapy/pyridostigmine) were little or no benefit. Novel use of oral salbutamol improved fatiguability, ptosis, oromotor dysfunction, muscle tone, articulation and voice volume in all patients. In 8/12 pregnancies maternal myasthenia gravis hadn't been established antenatally, and many mothers were pauci/asymptomatic. All had AChR-antibodies targeting the fetal γ -subunit confirming diagnosis. Where subsequent pregnancies were treated aggressively (immunotherapy), infants had improved outcomes.

Conclusions This report demonstrates and expands the phenotypic spectrum of FARIS, and emphasises oral salbutamol therapy as a potentially beneficial treatment. FARIS should be considered (mothers or infants tested for fetal specific AChR-Abs) in infants presenting with neonatal hypotonia, myopathic features and/or a suggestive antenatal history, even in the absence of a maternal MG diagnosis. Aggressive treatment with immunotherapy in pregnancy may improve outcomes.

GP231 REVIEW OF INVESTIGATIONS CARRIED OUT DURING THE FIRST PRESENTATION OF ACQUIRED DEMYELINATING SYNDROMES OVER A TEN YEAR PERIOD

Susan Harvey*, Niamh McSweeney. Cork University Hospital, Cork, Ireland

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Aims A first episode of suspected demyelination presents a diagnostic challenge often having non-specific signs which overlap with other inflammatory white matter, neurometabolic and genetic disorders. The first episode may be a presentation of acute disseminated encephalomyelitis, multiple