GP48 GERMAN NEWBORN SCREENING PROGRAMME IDENTIFIES FIRST CASE OF MCADD IN AN IRISH TRAVELLER CHILD IN THE ROI

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Aim/Purpose To report the first case of Medium-chain acyl-CoA dehydrogenase deficiency (MCADD) in an Irish Traveller child in the Republic of Ireland (ROI).

Methodology We describe the clinical presentation, outcome to date and genotype of a now 8 month old female infant from the Irish Traveller Community who was identified through newborn screening in Germany where she was born.

Results An 8 month old girl from the Irish Traveller Community returned to Ireland and was referred to local Paediatric services by her family doctor having been diagnosed with MCADD on the National Newborn Screening Programme in Germany.

Her mother had travelled to Germany for a wedding and developed placenta praevia. It was felt that it was too dangerous for her to travel back to Ireland at the time. The child was born at term by spontaneous vaginal delivery. She had a positive newborn screening test for MCADD. Subsequent metabolic tests confirmed this diagnosis.

On assessment at 8 months of age she was found to be robust, thriving with normal neurodevelopment. There have been no incidents of infection or hypoglycaemia. She has not required hospitalisation. Vaccines are up to date with no adverse incidents.

She was found to have the common p.(Lys329Glu), c.985A>G pathogenic mutation of the ACADM gene.

Conclusions This case highlights the success of the newborn screening programme in Germany and early medical intervention in MCADD. To our knowledge this is the first confirmed case of this recessive disorder in the highly consanguineous Irish Traveller community in the ROI.

GP49 MUNCHAUSEN BY PROXY SYNDROME IN THREE SIBLINGS DIAGNOSED AS ISOVALERIC ACIDEMIA

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Introduction Isovaleric acidemia (IVA) is a rare autosomal recessively inherited metabolic disorder caused by deficiency of isovaleryl-Co A dehydrogenase. Clinical signs are refusal of feeding, vomiting, lethargy, convulsions, metabolic acidosis, hyperglycemia/hypoglycemia, and foot odor in neonatal period. Munchausen by proxy syndrome is a complex form of childhood abuse that can be sometimes severe and fatal. A caregiver intentionally creates some symptoms mimicking an organic disease or changing the laboratory samples that causes damages in child health.

Case The patient who was diagnosed as IVA at 2-months of age had no other metabolic attacks until the age of seven. After that, he was hospitalized in every 3–4 months with severe dehydration and renal failure and hemodiafiltration was performed. The levels of creatinin and potassium are incompatible with life and there were any

sustaining clinical or electrocardiographical signs. His 2 year-old sister and 6 month-old brother who had also IVA were also hospitalized with recurrent encephalopathic status. As the family had a history of a child death without a known cause and the siblings had frequent hospitalizations in different medical centers and cities without a triggering factor, a more detailed anamnesis was obtained from the mother. Surprisingly this new history put forward the usage of antipsychotic/anatidepressive drugs in all siblings which lead to recurrent encephalopathies. However, renal insufficiency could not be explained until the mother's mixing of her urine to the blood samples of the child was observed in the clinic.

Conclusion Munchausen by proxy syndrome is a severe form of child abuse that can even result in death of the child if the diagnosis is not established in time. Although our patients had a known inherited metabolic disease with a defined mutation, the mother did not tell anything about this or sometimes told lies on the admissions to different medical centers. These confusing anamnesis easily lead the other physicians to the diagnosis of more common diseases like acute renal insufficiency, encephalities, and diabetes. Then, lots of unnecessary procedures was performed until the munchausen by proxy syndrome was thought. We present this case to emphasize thinking Munchausen by proxy syndrome with recurrent unexplained signs and symptoms even in the presence of a known disease in order to prevent undesirable results.

GP50 AUTOIMMUNE ENCEPHALITIS WITH SEVERE CATATONIA AND THYROTOXICOSIS

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Previously healthy 12-year-old girl. Ten days before presentation to our center presented focal seizures with secondary generalization. Studied in a local hospital, was started on levetiracetam. 4 days before transfer to our center she became disoriented with visual and auditory hallucinations, orolingual dyskinesias, sleep disturbances, enuresis and gait instability with no fever or other associated symptoms. She was suspected to have autoimmune encephalitis, received 1 gr/kg Immunoglobulins (IVIG), and transferred to our center.

At presentation, she was slightly tachycardic, inattentive, bradypsychic, with a decreased language fluency, disoriented in time and space, had orolingual dyskinesias, normal tone, and no nuchal rigidity or meningeal signs. No other relevant findings on physical exam.

Lumbar puncture showed leucocyte 20×uL, negative gram and culture, IgG 3.9 mg/dL (serum/LCR IgG index 0.43), IgG sintesis -10.8 mg/24 hrs, albumin 16.4 mg/dL (serum/LCR albumin index 3.57 normal), positive NMDAR antibody on LCR and positive oligoclonal bands.

Persistently tachycardic, with a TSH of 0.206 uUI/L, Free T4 2.82 ng/dL, T3 91.9 ng/dL, elevated Antiperoxidase antibody (113.88 UI/mL) and Anti Thyroglobulin (57.42 UI/mL) were compatible with thyrotoxicosis. Beta blockers were started with an adequate response.

She received a second dose of IVIG and 5 pulses of methylprednisolone but developed progressive neurological compromise, catatonia, and difficulty swallowing, requiring nasojunal tube feedings.

Plasmapheresis was initiated with a regular response. She required electroconvulsive therapy (ECT) for catatonia management. After the first ECT, she developed focal left frontal status epilepticus, was switched to phenytoin and valproic acid with good response. ECT was restarted after no crisis where detected, completing 8 sessions. Rituximab was initiated, completing four doses. There was a slow but steady recovery in her mental and neurological status, no signs of catatonia, persisted with an altered short memory, language difficulties, slightly inattentive, with emotional lability, recovered a normal gait and oral feedings. Paraneoplastic markers where all negative. She was discharged after 53 days of hospitalization with prednisone, valproic acid, Quetiapine, and lorazepam.

This patient presents a severe case of Autoimmune Encephalitis, with severe psychiatric symptoms and other autoimmune manifestations. Usually, there is a good response after treatment with IVIG and/or methylprednisolone. Rituximab was chosen to reduce de antibody production. Follow up as an outpatient, she has had no relapses and continues to recover.

GP51

PANDORAEA SPUTORUM IN A PAEDIATRIC CYSTIC FIBROSIS PATIENT

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Introduction Cystic fibrosis (CF) is the most frequent fatal genetic disease among caucasians. It is caused by mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) protein, which is found in all exocrine tissues, leading to thick, viscous secretions in several organs. The major cause of morbidity and mortality in CF's patients is lung function's deterioration due to pathogenic colonization with several bacteria. One emerging multidrug resistant pathogen that has been identified in CF is *Pandoraea sputorum*, a non-fermenting Gram negative bacteria.

Case report A 8-year-old boy, diagnosed with CF at 4 years old (homozygous genotype F508del; sweat test positive: 150 mEq/L) was admitted to the hospital for intravenous treatment of *Pandoraea sputorum* bronchopulmonary colonization. This pathogen was identified by matrix-assisted laser desorption ionization–time of flight mass spectrometry. He had a history of *Pseudomonas aeruginosa* lung colonization at 4 years old, and of *Stenotrophomonas maltophilia* at 7 years old, all of which had been eradicated after intravenous antibiotic treatment. All prior respiratory exacerbations were also treated empirically. His previous pulmonary function test revealed a forced expiratory volume in 1second (FEV1) of 89%. He was asymptomatic before, and throughout his inpatient stay, that lasted for 14 days while he received intravenous imipenem (80 mg/Kg per day). Two weeks after being discharged he was assessed at a paediatric pneumology appointment, remaining without symptoms, and with a FEV1

of 88%. His follow-up sputum culture was negative for *Pandoraea sputorum*.

Discussion In this clinical case the authors followed the protocol for early eradication, treating when the pathogen is first acquired, regardless of age or clinical signs or symptoms. This was based on several case reports that compared *Pandoraea sputorum* to *Pseudomonas aeruginosa* in terms of pathogenicity and decline of lung function. There is still a need to better understand this microorganism, and its correlation with disease outcomes, that can arise from the description of more case reports.

GP52

THE UNSUSPECTED HUMAN METAPNEUMOVIRUS RESPONSIBLE FOR A SEVERE ERYTHEMA MULTIFORME: AN UNUSUAL ASSOCIATION

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Erythema Multiforme (EM) is an acute immune-mediated condition characterized by the appearance of typical target-like lesions on the skin. They most commonly appear in a symmetrical distribution on the extensor surfaces of the acral extremities and subsequently spread in a centripetal way. EM 'major' involves oral, genital and ocular mucosae with erosions or bullae. Although cutaneous lesions are usually asymptomatic, EM can be caused by drugs, autoimmune disease, malignancy, irradiation, sarcoidosis and in 90 percent of cases by infections (viral, bacterial, fungal). Herpes Simplex virus is the most frequent etiologic agent. *Mycoplasma Pneumoniae* infection is another important cause of EM, particularly in children. Laboratory findings are not specific and clinical finding are necessary for diagnosis. A skin biopsy should be performed when the diagnosis is unclear. **CASE REPORT:** 14 year old male came to our attention for the appearance of cutaneous lesions, accompanied by high fever. The skin appeared almost entirely affected by roundish, sharp, erythematous lesions, some of these with evident 'coccard' sign, other ecchymotic with hemorrhagic nuance, confluent to the trunk in large patches. No recent history of infections or drugs. Laboratory findings showed a neutrophilia (N 8810/mcl) and eosinophilia (E 980/mcl) and high inflammatory indices (PCR 4.75 mg/dl, ferritin 517 kg/ml). Peripheral smear, autoimmunity, virological and bacterial screening and instrumental examinations were negative. On the third day of admission, he performed a nasal swab (Multiplex) due to the appearance of rhinorrhea and cough. It was positive for Human Metapneumovirus (HM). On the seventh day, there was a new poussé of erythematous, itchy, coccard element on the whole body surface. He was treated with antihistaminic, steroid and antibiotic therapy with gradual rash regression, desquamation of skin lesions and defervescence. In literature it is known that HM is a common cause of upper respiratory tract infection in children. However, no further cases are reported regarding the possible relationship between skin lesions and HM. In our case the only laboratory finding associate to the EM was a positive RT-PCR for HM. This observation could lead to further scientific evaluations.