segregation error of maternal reciprocal translocation t(9;13) (p21;q12).

Conclusion Balanced reciprocal translocations in either parents can amplify and produce unbalanced gametes leading to defective conceptus. Prenatal diagnosis is strongly recommended where balanced translocation is found in parent. Clinical features of the affected conceptus depends largely on the regions of chromosome involved.

P573 A CASE OF INSIDIOUS RECURRENT ABDOMINAL PAIN

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Porphyrias are a group of inherited metabolic disorders of heme biosynthesis leading to excessive accumulation and excretion of porphyrins. The variable clinical manifestations may determine a delay in the diagnosis, followed by a possible negative clinical outcome.

We describe a case of a 14-year-old girl with Shwachman-Diamond syndrome (SDS) and epilepsy admitted with severe cyclic abdominal pain not responsive to antalgic therapy, localized in the epigastric area, irradiating to the whole abdomen and the back, associated to aspecific symptoms (diarrhea, fever, vomiting). During hospitalization, the girl was asymptomatic. We excluded infectious, autoimmune, endocrinological causes, a relapse of SDS and performed a screening for porphyria that resulted negative. After each attack, she referred presenting urinary retention followed by hyperchromic urines, anxiety and paresthesias. Past clinical data showed hyponatremia, tachycardia and hypertension. Eventually, increased values of ALA (8.21 mg/l) and PBG (3.96 mg/l) were found; exposure of fresh urines to sunlight caused a change of their color. Finally, genetic analysis was negative. These findings allowed the diagnosis of porphyria, therefore we prescribed a normocaloric hyperglucidic diet and preventive glucose solutions in stressing situations, with regression of symptoms.

Acute porphyrias present with life-threatening crisis secondary to the injury of central, peripheral and autonomic nervous system. They can be triggered by drugs, alcohol, infections, reduced caloric intake, endogenous hormone cycles and stressing situations. The characteristic manifestations are severe cyclic abdominal pain, neurological or psychiatric symptoms and/or hyponatremia. Diagnostic is the assessment of plasmatic PBG/ALA, always increased during an attack, normal during remission. Treatment is human haemin although, in mild attacks, a diet with high carbohydrates and/or preventive glucose infusions is effective; a negative genetic evaluation should never exclude the diagnosis.

It is important considering acute porphyria in the differential diagnosis of severe cyclic abdominal pain, particularly in subjects affected by different and/or rare clinical disorders.

P574 COMPLEX REGIONAL PAIN SYNDROME AND HPV VACCINE: A CASE REPORT OF COMPLEX REGIONAL PAIN SYNDROME AND LITERATURE REVIEW

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Introduction Complex Regional Pain Syndrome (CRPS) is a chronic pain disorder characterised by significant autonomic features which typically develops in an extremity after acute tissue trauma. Symptoms may include continuing pain, sensory abnormalities like allodynia, hyperalgesia, hypoesthesia, vasomotor abnormalities in skin colour or temperature, sudomotor abnormalities of sweating or oedema and motor/trophic abnormalities in hair, skin, nails, tremor or muscle weakness. HPV vaccination is offered to all girls in Northern Ireland aged 14/15 years and recent plans have been made to offer it to boys starting late 2019. Since the introduction of HPV vaccination there have been case reports across the world describing various side effects after HPV vaccination, including Complex Regional Pain Syndrome. One report alone detailed 45 patients across 13 countries. In Japan the vaccination was temporarily halted as a consequence. The Vaccine Adverse Event Reporting System (VAERS) USA, has received 31,911 reports of adverse events following any HPV vaccination from which 22 (0.07%) reports met the criteria for complex regional pain syndrome (including reflex sympathetic dystrophy) as an adverse event. 21 cases were after Gardasil (HPV4) vaccine and 1 was after Cervarix (HPV2) vaccine.

Case We report a previously fit healthy 14 years old Irish girl. She had received her 2nd HPV (Gardasil) vaccine on 09/04/18 having received her 1st vaccine without any problems in October 2017. Twenty minutes post vaccine her left arm from the elbow down became swollen, purple and painful. Presentation was consistent, CRPS which was initially not diagnosed at first presentation to ED and Paediatrics. Correct diagnosis made four weeks post event and appropriate treatment initiated with full recovery within eight months.

Laboratory investigations No significant abnormalities.

Conclusion There are documented cases of CRPS after HPV vaccine although is still controversial as to whether it is a vaccine or the effect of the trauma associated with inserting a needle which is the aetiological factor. This case highlights the need for better recognition of this condition amongst paediatricians as earlier initiation of treatment is associated with better long term outcome.

P575 CASE REPORT: TWINS WITH NIEMAN PICK SYNDROME

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Introduction Niemann-Pick Disease is an autosomal recessive metabolic disorder characterized by deposition of sphingomyelin and cholesterol within the lysosome of reticuloendothelial cells of various organs due to inherited deficiency of an enzyme, acid sphingomyelinase. Niemann-Pick Disease is classified into four types such as A, B, C and D each caused by a different gene mutation.

Case We report a case of Niemann Pick Disease type A/B twins. The patients are 3 years old girls, who presented to Sligo University Hospital with repeated episodes of chest

infection. On examination they were found to have hepatosplenomegaly, with normal cognition and development. Eye examination was normal, basal blood tests were normal as well as metabolic screen and cardiac Echo, CT chest showed interstitial changes, lysosomal enzyme screen showed marked deficiency of (ASM) Sphingomyelinase activity and increased Chitotriosidase activity which is consistent with Nieman pick Type A/B disease. A blood sample has been sent for full gene sequencing of the Nieman Pick A/B gene. Currently they are well and thriving, with no major complications in the mean time

Discussion There is no specific treatment for NPD. Orthotopic liver transplantation in an infant with type A disease and cord blood transplantation in several type B NPD patients has been attempted with little or no success. BMT in a small number of type B NPD patients has been successful in reducing the spleen and liver volumes, the sphingomyelin content of the liver, the number of Niemann-Pick cells in the marrow, and radiologically detected infiltration of the lungs. ERT with recombinant human ASM is currently in clinical trials for the treatment of type B patients

Conclusion Niemann-Pick disease types A and B is estimated to affect 1 in 250,000 individuals, Chronic visceral ASMD could have a mild course with a relatively good outcome. Other factors such as environmental ones could contribute to the disease severity.

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KERION: A NASTY SCALP INFECTION

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Background/Aims Kerion is a scalp condition that occurs in severe cases of scalp ringworm (tinea capitis) with the highest prevalence in children 3 to 10 years of age. It appears as an inflamed, thickened, pus-filled area, with scaly spots or patches of broken hair on the scalp. A kerion is treated with oral antifungal medicines because the fungus grows deep into the hair follicle where topical creams and lotions cannot penetrate. Oral corticosteroids are also started in cases where lesions are tender and to reduce the inflammation. Kerion is usually associated with infection by zoophilic dermatophytes, Tricophyton Verrucosum and Tricophyton Mentagrophyte, although other sources have been described. Our aim is to report an unusual scalp infection.

Methods The patient history, clinical presentation with photographs, examination and laboratory findings (skin scrapings were taken for mycology), treatment and outcome are described.

Results A previously well 6-year-old boy, of African origin, presented to the Paediatric Emergency Department with a 6-month history of scalp infection. It initially started on the right temporal region and was treated with Canesten cream for 6 months. There was no improvement and the condition was treated with Flucloxacillin a week prior to presentation to us, at which point it had worsened and started spreading to the rest of his scalp. On examination there was a raised demarcated lesion on his right temporal area which appeared crusted and discharge was noted. There were also multiple round, dry, scaly patches on his scalp which were not infected. The dermatology team commenced him on oral

Itraconazole, Nizoral Shampoo and Paraffin Gel for 6 weeks, and oral steroids for 1 week.

Conclusion Kerion is not often seen in our paediatric department. This case raises awareness of a rarely seen condition in an Irish context.

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JOINT MANIFESTATIONS AS A FORM OF ONSET IN CROHN'S DISEASE

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Crohn's disease (CD) is an inflammatory bowel disease (IBD) of multifactorial etiology that affects all segments of the gastrointestinal tract and is associated with multiple extra-intestinal manifestations. Spondyloarthropathies are the most common extra-digestive manifestation of IBD's. Spondyloarthropathies primarily involve the axial skeleton, but may also be associated with peripheral symptoms such as synovitis, dactylitis or enthesitis.

Objectives Case presentation of a teenager, aged 14, diagnosed with CD A1 L3L4B1p-score PCDAI 45, (November 2018) that presented with joint manifestations as a form of onset.

Case presentation The patient exhibits joint pain at the level of large joints of self-limiting character, as well as pain at the level of the costochondral joints associated with fever spikes. In recent history, over a period of two months, the patient presented up to 10 stools/day, without pathological features, with consecutive weight loss. An infectious etiology was suspected and empiric antibiotic treatment was instituted, with the evolution being unfavorable.

She is admitted to the clinic for further investigation and treatment. Biologically she presented: marked inflammatory syndrome, reactive thrombocytosis, hypochromic microcytic anemia, positive calprotectin. The suspicion of an IBD, is raised, multiple biopsies from the upper digestive tract being performed. The macro and microscopic appearance suggested Crohn's disease in the stage of active lesions. Induction therapy with exclusive enteral nutrition (EEN) is established, as well as azathioprine, to maintain remission. Evolution is favorable with the remission of joint symptoms in the first 2 weeks of EEN.

Conclusion In some cases, patients with Crohn's disease, due to chronic inflammatory syndrome, may develop symptoms similar to those encountered in spondyloarthropathies.

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INTRAVENOUS MAGNESIUM SULPHATE INFUSION IN THE MANAGEMENT OF SEVERE TETANUS: A CASE SERIES

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Background Tetanus is still prevalent in developing countries. In the management of tetanus, eradication of bacteria from the wound and neutralization of remaining toxins are detrimental. However, supportive management especially control of muscle spasm and airway protection is equally important. We