Hypomelanosis of Ito, a rare neurocutaneous syndrome associated with muscular, skeletal, neurological alteration and ophthalmologic abnormalities.

Methods A 9 month old girl was brought for evaluation of hypopigmented skin lesions and strabismus. Clinical examination revealed whirled hypochromic skin lesions only in the left side of the body (neck and trunk) following the lines of Blaschko, compatible with HI and hypopigmentation. She presents also: strabismus, genu valgus and pubis pilosity. She attained head control at 4 months and failure to sit without support.

Results The blood tests, hormonal tests, abdominal and cranial ultrasound were normal. Electroencephalogram revealed diffuse cortico-subcortical dysfunction. MRI of head was normal. Blood karyotyping showed mosaicism. Differential diagnosis: nevus cortico-subcortical dysfunction. MRI of head was normal. Blood and hormonal tests were normal. The levels of breast milk Hg levels were higher in infants with increased family, living in older house and in urban with air pollution at the 8th month were inversely correlated with z-scores of birth weight, height for age at the 8th month were inversely correlated with infant hair Pb levels. In boys, z scores of body mass index at the 8th month were inversely correlated with infant hair Cd levels. The levels of breast milk Hg levels were higher in infants with increased family and impaired anaemia. The authors report a case of Hypomelanosis of Ito (HI), a rare neurocutaneous syndrome associated with muscular, skeletal, neurological alteration and ophthalmologic abnormalities.

Methods and description The examination of the Mucopolysaccharidosis type I (MPS I) indicate hyperplastic gingiva, macroGLOSSIA, high-arched palate, short mandibular rami with abnormal condyles, spaced hypoplastic peg-shaped teeth with retarded eruption; and localised dentigerous cyst-like radiolucencies. Guven et al. (2008) have investigated the ultra structural and chemical properties of MPS I (Hurler) teeth. The dentin of the primary teeth was characterised by extremely narrow dentinal tubules with an irregular wave-like pattern. The enamel-dentin junction was poorly shaped, micro gaps occurred and the enamel displayed an irregular arrangement of prisms. The enamel and the dentin had an abnormal protein structure and the dentin protein content was low. The mucopolysaccharidoses (MPS) are prominent among the lysosomal storage diseases. The intra-lysosomal accumulation of glycosaminoglycans (GAGs) in this group of diseases induces a cascade of responses affecting cellular functions and maintenance of the extra-cellular matrix. As well as skeletal problems, mucopolysaccharidoses’ patients have dental with specific deformities. Teeth involvement is highlighted, having an eye to the possibilities of reversing these oro-dental changes with enzyme replacement therapy.

A novel case of unbalanced translocation of chromosome 3 and 7

Background It is estimated that 1 in 500 to 1 in 625 human newborns have a balanced reciprocal chromosomal translocations. Such individuals are usually healthy and do not have any specific features. We report an unusual case of unbalanced translocation of chromosome 3 and 7 and describe its features.

Case report A female baby born at 37 weeks gestation by a spontaneous vaginal delivery was admitted to our NICU with poor Apgar scores. Immediately after birth, she had a poor ventilatory effort requiring continuous positive airway pressure (CPAP) and further deterioration in her respiratory function led to intubation and surfactant administration. Echocardiogram revealed small perimembranous VSD. She had dysmorphic features including
micrognathia, broad occiput, low set ears, single palmar crease, and large cleft palate. Subsequent genetic tests confirmed unbalanced translocation of chromosome 3 and 7. She was gradually weaned off ventilator support at 2 weeks and was discharged from NICU. A week following discharge she presented with bronchiolitis and has continued need for High flow (Vapotherm) support. She feeds on high energy formula via a nasogastric tube due to poor weight gain and remains on treatment for moderate to severe reflux. She is likely to need gastrostomy and cleft surgery.

Conclusion Dysmorphic features as reported in this case report should raise suspicion of a chromosomal defect, which needs early genetic referral and microarray. Balanced translocations are common and usually do not have specific clinical features. However, unbalanced translocations are uncommon but they may have significant clinical expressions.

PO-0378 SAFETY OF BIFIDOBACTERIUM ANIMALIS SUBSP. LACTIS (B. LACTIS) STRAIN BB-12-SUPPLEMENTED YOGHOURT IN HEALTHY CHILDREN: A PHASE I SAFETY STUDY

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Probiotics are live microorganisms that, when administered in sufficient doses, provide health benefits on the host. The purpose of the study is to determine the safety of Bifidobacterium animalis subsp. lactis (B. lactis) strain BB-12 (BB-12)-supplemented yoghurt when consumed by generally healthy children. Secondary aims are to evaluate the influence of BB-12 on the faecal microbiome and changes in the microbial community. A phase I, double-blinded, randomised, placebo-controlled study was conducted in compliance with United States Food and Drug Administration guidelines for an Investigational New Drug (IND). Sixty participants were randomly assigned to consume four-ounces of the active yoghurt supplemented with BB-12 or placebo yoghurt daily for 10 days. The primary outcome was to assess safety and tolerability, assessed by the number of reported adverse events. Preliminary results show 181 non-serious adverse events were reported, with no differences between the groups. Three serious adverse events unrelated to the yoghurt interventions were reported. BB-12 supplemented yoghurt is safe and well-tolerated when consumed by healthy children. Faecal samples collected before, during and after the intervention period will be analysed using state-of-the-art DNA sequencing and analysis tools to assess the relationship between the microbiome and probiotics, and to provide novel information on the dynamics of the complex ecosystem in the human gut. This study will form the basis for future clinical trials investigating the potential effects of BB-12 supplemented yoghurt in a variety of disease states.

PO-0379 DIFFERENT ASPECT OF CHILDHOOD LANGHERHANS CELL HISTIOCYTOSIS : EXPERIENCE FROM A SINGLE CENTRE

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Introduction Langerhans cell histiocytosis (LCH) is characterised by a reactive clonal proliferation and accumulation of dendritic cells with a wide range of clinical presentations.

Survival rate depend on single or multisystem disease.

Objectives The aim of the study was to analyse the clinical, radiologic features and responses to treatment.

Materials and methods We retrospectively reviewed the clinical data, histopathological, radiologic features, treatment modalities, and outcome of patients presenting with LCH.

Results 9 patients were included with two brothers. There were 5 girls and 5 boys. Mean Age at diagnosis was 39 months. The main clinical feature was prolonged fever (5 cases), and impaired general condition (3 cases).

Skin involvement was present in 5 patients, otitis in 3 patients and 3 cases of lung injury with Spontaneous pneumothorax in one case. 3 different Tumour syndromes were observed at diagnosis. The most of patients present a multi-system disease.

Radiologic finding showed 2 cases of bone involvement. The bone marrow involvement was present in 2 patients. Six patients received corticosteroid and vinblastine combination with the use of cyclosporine in 3 cases. One patient developed insulin diabetes. Two patients dead.

Conclusion Childhood Langherans cell histiocytosis is a rare and poorly understood multi-system disease. Treatment decisions are difficult given the unpredictable course of the disease sometimes spontaneous, mainly for unifocal forms remissions.

Patients with localised disease generally have a good prognosis and require minimal treatment. However, patients with lesions in ‘risk’ organs (liver, spleen, lung, bone marrow) have a worse overall prognosis regarding mortality and morbidity.