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 islikely 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PO-0377 WITHDRAWN

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

[1]DJ Merenstein, [1]TP Tan, [1]KA Herbin Smith, [2]RF Roberts, [3]FJ D'Amico. *[1]Department of Family Medicine, Georgetown University Medical Center, Washington DC, USA; [2]Department of Food Science, The Pennsylvania State University, University Park PA, USA; [3]Department of Mathematics and Computer Science/St. Margaret Family Medicine Residency Program, Duquesne University/University of Pittsburgh Medical Center, Pittsburgh PA, USA*

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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 yoghourt 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 yoghourt supplemented with BB-12 or placebo yoghourt 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 yoghourt interventions were reported. BB-12 supplemented yoghourt 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 yoghourt in a variety of disease states.

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

[1]M Tfifha, [1]O Gasmi, [1]H Ajmi, [1]S Mabrouk, [2]K Kadri, [1]J Chemli, [1]N Zouari, [1]S Hassayoun, [1]S Abroug. *[1]Pediatrics, University Hospital Sahloul, Sousse, Tunisia; [2]Radiology, University Hospital Sahloul, Sousse, Tunisia*

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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 ferver (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 insipidus diabetes. Two patients dead.

Conclusion Childhood Langherhans 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.

PO-0380 PARENTAL EMOTIONAL REACTIONS TO PAEDIATRIC HOSPITALISATION

P Tsitsani, P Stefanakaki, V Blogoura, V Moschou. *Paediatric Clinic, General Hospital of Didimoteixo, Didimoteixo, Greece*

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Background and aims When the child is admitted to hospital, parents are faced with a crisis generated by the traumatic events of disease and hospitalisation. This questionnaire-based survey aims to explore the emotional responses of parents to their children's hospitalisation and associate them to health-care professionals' behaviour.

Methods Parents (80 mothers, 75 fathers) of 155 children aged 2–12 years, hospitalised at least 4 days, completed the questionnaire and engaged in private structured interviews. The questionnaire contained demographic data (age, sex, and educational background), questions about parent's psychological characteristics in general and their specific emotions during the pre-admission and in-hospital phase.

Results Participants were 10% affluent, 65% from middle-class social groups and 25% of relatively deprived minority status. Two – thirds of parents regard themselves as overprotective and highly anxious, 32% present responsible and caring and 1% tend to be indifferent. 19% of parents initially refused admission. The major pre-admission emotions are self- blame, anger and fear, positively associated to previous hospital stays. Stress and discomfort dominate through the in-hospital phase, during which 23% of parents experienced psychosomatic symptoms. Negative emotions become reversed when the need for information by doctors and nurses is satisfied. Fathers expressed less effectively both positive and negative emotions.
Limitations Parents of children with chronic diseases were excluded from the study.
Conclusions Children's sickness and hospitalisation disorganizes the family, resulting in parental emotional disturbance, confusion and uncertainty. Stress-related emotions are better coped with, when parents feel they are respectfully treated and adequately fed-back by the hospital personnel.

PO-0381 KARTAGENER SYNDROME ASSOCIATED TO NEPHROPATHY: A BROADER SPECTRUM OF CILIOPATHY?

[1]E Veiga, [1]A Veiga, [1]J Llerena Junior, [1]C Serao, [1]C Mochdece, [2]P Oliveira Goncalves, [2]S Teixeira, [2]R Semenzim. *[1]Pediatria, Faculdade Medicina de Petrópolis, Petrópolis, Brazil; [2]Graduação Em Medicina, Faculdade Medicina de Petrópolis, Petrópolis, Brazil*

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Introduction Kartagener Syndrome (KS), an autosomal recessive disorder, subgroup of primary ciliary dyskinesia, is characterised by situs inversus, bronchiectasis and chronic sinusitis.
Objective To report a case of KS and associated nephropathy, diagnosed at 6 years in Hospital Alcides Carneiro (HEAC).
Method Descriptive and observational evolution of MESF, hospitalised in the paediatric ward of the HEAC, with abdominal pain and nephritic proteinuria. Clinical diagnosis of SK was established only at 6 years, despite dextrocardia diagnosed at 3 months and several subsequent hospitalizations due to respiratory infections. Nephritis was identified at 4 years, slowly progressive and without specific etiological cause determinated. Tomography showed bronchiectasis and *situs inversus totallis*. No identifiable chromosomal abnormalities seen in conventional karyotyping and no molecular study could be performed. Clinical diagnosis, however, made possible hearing loss and lung infections prophylaxis and genetic counselling. Although there are some reports of genitourinary abnormalities associated with SK, it's important to determine if KS is the specific cause of nephropathy, due the intimate relationship of other ciliopathys with renal disease, as observed in Bardet-Biedl syndrome and Meckel syndrome. In this patient the aetiology of nephropathy is unclear so far, and most common causes have been discarded.
Conclusion Factors such as low incidence estimated at 1:17000 births and pathophysiological complexity are considered contributory to late diagnosis, worsening of pulmonary disease and consequently worse prognosis. We couldn't perform molecular diagnosis in the several genes related to SK, which could provide preimplantation diagnosis and establish the best genotype-phenotype, especially in correlation with nephropathy.

PO-0382 DEVELOPMENTAL DELAY IN TRISOMY 21 CHILDREN FROM ROMANIA

[1]RM Vlad, [2]P Grigorescu-Sido, [2]S Bucerzan, [2]C Al-Khzouz, [2]I Nascu, [1]D Oraseanu. *[1]Pediatrics, "Grigore Alexandrescu" Emergency Children's Hospital "Carol Davila" University of Medicine and Pharmacy, Bucharest, Romania; [2]Medical Genetics, "Iuliu Hatieganu" University of Medicine and Pharmacy, Cluj Napoca, Romania*

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Aims Trisomy 21 is the most frequent autosomal chromosomopathy. The developmental delay is a constant finding. This study aimed to evaluate the auxological parameters of Romanian trisomy 21 patients and to correlate them with the cytogenetic findings.
Material and method We conducted an observational study on 136 patients with cytogenetically confirmed trisomy 21 that were evaluated recording their auxological parameters. The nutritional status of the patients was evaluated using growth charts from WHO until 5 years and from CDC over 5 years. For infants, the ponderal index was used. For patients ≥1 year, we used growth charts for weight and BMI and standard deviation score for height. The growth parameters were statistically evaluated considering three age groups (newborn, infant, child ≥1 year) and the cytogenetic anomalies.
Results 91.2% of patients had regular trisomy 21. The mean birth weight was 2904grams and the mean birth length 49,6cm. 80% of the infants associated protein-caloric malnutrition; the length was affected in 54.9% of 1 month - 1 year patients. 49.4% of patients ≥1 year were underweight, 37.03% were overweight and 75.3% associated short-stature. Only 14.5% of patients <1 year and 8.6% of patients ≥1 year had a normal nutritional status. No significant differences were found in relation to the cytogenetic diagnosis.
Conclusions The somatic delay varied in relation to age. In infants underweight dominated. In children ≥1 year short-stature was the main finding and one third of patients were overweight. Only a reduced number of trisomy 21 children had normal auxological parameters.

PO-0383 MONITORING IGG ON ORAL FLUID FOR THE MANAGEMENT OF CHILDREN AT RISK FOR CONGENITAL TOXOPLASMOSIS

[1]M Wallon, [2]E Chapey, [3]V Meroni, [4]F Kieffer, [5]P Garcia-Meric, [6]C L'Ollivier, [1]F Peyron. *[1]Institut de Parasitologie Et de Mycologie Médicale, Hôpital de La Croix-Rousse, Lyon, France; [2]Faculté de Médecine Et de Maïeutique Lyon Sud, UCBL, Lyon, France; [3]Fondazione IRCCS, Policlino San Matteo, Pavia, Italy; [4]Service de Néonatalogie, Hôpital Trousseau, Paris, France; [5]Service de Médecine Néonatale, Hôpital de La Conception, Marseille, France; [6]Laboratoire de Parasitologie Mycologie Médicale, Hôpital de La Timone, Marseille, France*

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Background and aims Despite progress made for the diagnosis of congenital *Toxoplasma* infection *in utero* and at birth, serological follow-up in first year of life remains required to exclude or confirm congenital infection. To reduce the constraints of this follow-up, we developed a test to detect anti-*Toxoplasma* IgG in oral fluid.
Methods In 362 patients referred for *Toxoplasma* serology oral fluid was collected on two micro-sponges in parallel to blood sampling. A pilot study on 212 patients aged >15 months (274 samples) was performed to validate sampling procedures and develop an in-house indirect ELISA for the detection of