Diagnostic was suspected on hypotonia with poor suck in the neonatal period in the first case, hypotonia with history of feeding difficulty and psychomotor developmental delay in the second case and hyperphagia with obesity in the third case.

Physical exam showed facial dysmorphy in 1 case, bilateral cryptorchidism in the 3 cases and obesity (BMI = 34.3) in the third case.

Chromosome analysis with fluorescence *in situ* hybridization (FISH) confirmed the diagnosis with identification of the deletion 15q11.2 – q13 in the three cases.

The average retreat was 2 years; the evolution was marked by morbid obesity (BMI=57) with hypertension and psychiatric disturbance with hyperactivity in the third case and significant weight gain at the age of 10 months in the second case.

Conclusion Prader Willi must be suspected in all newborns with unexplained persistent hypotonia and confirmed by chromosome analysis. Early diagnosis is important to effective long-term management.

PO-0367 | **FORMULA INTOLERANCE AND ORTHODOX DIETARY LAWS – A PASSOVER CONNEXION**

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Background When neonates and infants are not breastfed or require supplemental feedings, American Academy of Paediatrics recommends iron-supplemented formula. Cow's milk-based formula can be allergenic; however, this allergic response may be secondary to formula components as opposed to milk. The allergen may be inscrutable leading to false assumptions. In this case, an unusual religious clue solved such mystery.

Method Case analysis

Results An orthodox Jewish infant initially breastfed was transitioned to formula. Enfamil 4ounces q4hours caused upper respiratory mucus build-up directly post 'bottle-time'; formula was changed to ProSobee (soy-based) with resolution of 'formula intolerance.' At 12 months, cow's milk was initiated without allergic response. At 15 months, when diet included solid foods, she developed a peri-oral rash temporally associated with meals in her parents' home and following a meal in another family's home. Initial assessment revealed no new foods during these meals; however, this new allergy occurred during Passover, with strict dietary laws, suggesting a common new food. Ashkenazi Passover dietary laws preclude kitniyot (legumes, derivatives and associated crops including corn/soybeans/corn oil/soybean oil). Common Passover cooking oils include safflower oil. As both families were Ashkenazi Jews, the apparent common new food was safflower oil used in both homes. Safflower oil is an Enfamil ingredient. The presumptive allergen common to formula and Passover meals was safflower oil.

Conclusions Allergen determination may be secondary to fortuitous double exposure. Complete clinical histories need to allow for possible double exposure with willingness to consider cultural, religious, dietary law, and holiday dietary law factors.

PO-0368 | **WITHDRAWN**

PO-0369 | **A NIEMANN-PICK DISEASE TYPE C (NP-C) SUSPICION INDEX TOOL TO AID DIAGNOSIS IN PAEDIATRIC PATIENTS**

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NP-C is a lysosomal lipid storage disorder caused by mutations in *NPC1 or NPC2* genes. NP-C can present with a range of visceral, neurological and psychiatric symptoms that vary with age. A suspicion index (SI) tool was developed to assist clinicians achieve early diagnosis. The tool accurately predicts NP-C in patients >4 years of age but performs poorly in paediatric (≤4 years) patients. The present study aimed to utilise the characteristic symptomatology of NP-C in paediatric patients to develop a novel tool to assist paediatricians to identify patients for NP-C testing.

Paediatric patients were classified according to diagnosis: NP-C suspected and confirmed (n = 106); NP-C suspected but negative (n = 31); control (no suspicion of NP-C; n = 63). Symptomatology data were collected retrospectively by questionnaire and summarised descriptively. The relationships between individual symptoms and likelihood of confirmed diagnosis of NP-C were defined by statistical modelling. The final tool was developed iteratively using combinations of symptoms until optimal discriminatory power was achieved.

The characteristic symptomatology of paediatric NP-C patients was identified; visceral symptoms were more prominent compared with older patients. The new tool discriminates well between NP-C confirmed, NP-C negative and control subjects. Statistical analysis demonstrates superior sensitivity and specificity of the paediatric tool compared to the original tool. The newly developed paediatric NP-C SI tool will help paediatricians to identify more paediatric patients with a high suspicion of NP-C, leading to more referrals for specialist testing thus improving early diagnosis and management of NPC-disease in paediatric patients.

PO-0370 | **GERM CELLS INDUCED FROM HUMAN UMBILICAL CORD MESENCHYMAL CELL-DERIVED INDUCED PLURIPOTENT STEM CELLS BY BMP4**

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Objective To reprogramme the induced pluripotent stem (iPS) cells from Human umbilical cord mesenchymal cells (HuMSCs) and induce the iPS cells into germ cells by BMP4.

Methods OCT4, SOX2, Klf4,c-myc, Nanog, Lin28 were transfected into HuMSCs with lentivirus to reprogram HuMSCs into iPS cells. Morphological observation, Alkaline Phosphatase staining, karyotype analysis, RT-PCR, immunofluorescence staining, tumour formation *in vivo* and embryniod body formation *in vitro* were performed to examine the pluripotency of the iPS cell lines. Then we induced one of the iPS cells lines into germ cells by BMP4. Gene expression was measured by qRT-PCR at days 0, 3, 7, 10 and 14. Early-stage germ specific protein VASA and meiosis specific protein SYCP3 were assesssed by immunofluorescence staining.

Result We obtained two iPS cell lines completely reprogrammed, HuMSC-iPS1 and HuMSC-iPS2. HuMSC-iPS1 expresses germ cell markers at undifferentiated state. BMP4 can upregulate germ cell markers at different time points highly while the spontaneous differentiation just upregulate DPPA3, DAZL and VASA modestly at day 3. However, all of these genes were downregulated at day 14. VASA and SYCP3 immunofluorescence staining indicates there is a high VASA expression in BMP4 induced group in contrast to low expression in the spontaneous group at day 7. Meanwhile, there is a modest SYCP3 fluorescence in BMP4 induced group in contrast to no immunofluoresence in the spontaneous group.

Conclusion This system can reprogram HuMSCs into iPS cells effectively. The MSC- iPS1 can differentiate into early germ cells spontaneously while the germ cells induced by BMP4 can enter meiosis.

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Introduction Chronic recurrent multifocal osteomyelitis (CRMO) is a rare auto-inflamatory bone disease, primarily affecting children and adolescents.

Objective To determine the clinical features and functional outcomes of children with CRMO treated between 1995 and 2013 in a tertiary paediatric hospital.

Methods We retrospectively reviewed clinical features of children with CRMO. Outcomes were obtained through telephonic interview. The Health Assessment Questionnaire (HAQ) disability índex and the Quick Dash questionnaire were applied. Upper limb disability of patients with clavicular involvement was compared with a control group of children who had a previous forearm fracture.

Results Nine patients were assessed (6 males and 3 females). Mean age at onset was 8 years; mean age at diagnosis 10,3 years. Mean number of bony lesions were 3 at onset and 3,4 over the disease course. The more frequently affected site was clavicle (55,6%). Pain was the most frequent clinical sign of the disease (88,9%) associated with swelling (77,8%). Patients were treated with steroids (88,9%), methotrexate (44,4%), pamidronate (22,2%) and NSAID (100%). Five patients presented residual bone deformity (including two with inactive disease). All patients reported no physical disability as judged by the HAQ 0–1. However, patients with clavicular involvement (5) scored significantly worst on Quick Dash when compared to the control group (p = 0,015).

Conclusions Although the clinical outcome of children with CRMO is generally considered good, using an upper-extremity outcome measure (Quick DASH) allows us to understand that these patients experience upper limb difficulties in their daily lives.

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The clinical spectrum of Autoinmune hepatitis (AH) is very wide, ranging from asymptomatic individuals with abnormal liver function to those with fulminant liver failure. The objective of this study was to analyse the clinical presentation, biochemical and histological diagnosis of a number of cases of AH.

Methods between January-2006 and November-2013, we found 4 patients who were diagnosed AH.

Results The median age was 11.9 years (2–13), predominantly female (75%). The level of GOT and GPT were elevated, with a median of 259 IU (128–2451) and 309 IU (117–1449) respectively. The mean serum albumin was 3.9 ± 0.3 g/dl and the presence of IgG mean was 2391.5 ± 628. In all cases, the liver biopsy was performed to establish diagnosis. The anti-liver Kidney microsome type-1 (anti-LKM1) antibodies were present in all patients, and only in two of patients were found positive anti-smooth muscle (ASMA) antibodies. In 50% were positive for antinuclear (ANA). Among the associated comorbidities found a case of hypothyroidism and vitiligo. Note that two of the four cases correspond to the same family, where molecular genetic study was carried out, with both sisters carrying HLA haplotypes DRB1 07. DRB1 14/DQB1 02. DQB105. In the majority of patients had non-specific symptoms, and only one patient presented jaundiced with abdominal pain. At follow-up, all patients are alive and there were not progression to cirrhosis or fulminant liver failure

Conclusion In our series, the autoimmune hepatitis was characterised predominant in female, with non-specific symptoms, had levels of transaminases elevated and associated with other autoimmune pathologies.

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Aim Lead (Pb), mercury (Hg), cadmium (Cd) are found widely in environment and stay in nature for a long time. These toxic chemicals have some hazardous effects on child health. The purpose of this study are to determine the levels of heavy metals (Pb, Hg, Cd) in breast milk and infant hair, related