

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 dysmorphism 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.

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

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