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

OM Lourenc, M Pineda, E Mengel, B Heron, J Imrie, SA Jones, V van der Linden, H Jahnova, J Jesna, P Katritzedeh, Yi Vlagangianopoulos, JM Torres, Kolb.

1 Clinical Genetics, University of São Paulo, São Paulo, Brazil; 2 Fundación Hospital Sant Joan de Déu CIBERER, Instituto de Salud Carlos III, Barcelona, Spain; 3 Villa Metabolica, ZKIM MC University Mainz, Mainz, Germany; 4 Centre Référence Des Maladies Lysosomales, CHU Trousseau APHP, Paris, France; 5 Central Manchester and Manchester Children’s Foundation Trust, University of Manchester, Manchester, UK; 6 Manchester Centre for Genomic Medicine, Central Manchester University Hospitals Foundation Trust, Manchester, UK; 7 Paediatric Association for Assistance to Disabled Children (AADD); Pernambuco, Brazil; 8 Institute of Inherited Metabolic Disorders, Charles University, Prague, Czech Republic; 9 Department of Pediatrics and Adolescent Medicine, Charles University, Prague, Czech Republic; 10 Pediatric Research Center, Shahid Beheshti University of Medical Sciences, Tehran, Iran; 11 Centre Référence Des Maladies Héréditaires Du Métabolisme de l’Enfant Et de l’Adulète, Hôpital Universitaire Necker-Enfants Malades, Paris, France; 12 Biostatistics, Syntax for Science SL, Basel, Switzerland; 13 Global Medical Lead, Actelion Pharmaceuticals Ltd, Allschwil, Switzerland

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.

Supported by Actelion Pharmaceuticals Ltd.

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

1 W, 2 Y Wang, 3 Y Chen. 1 Pediatrics, The Second Affiliated Hospital of Shantou University Medical College, Shantou, China; 2 Pediatrics, Capital Institute of Pediatrics, Beijing, China; 3 Pediatrics, Guangdong Maternity and Child Health Care Hospital, Guangzhou, China

10.1136/archdischild-2014-307384.1017

GERM CELLS INDUCED FROM HUMAN UMBILICAL CORD MESENCHYMAL CELL-DERIVED INDUCED PLURIPOTENT STEM CELLS BY BMP4

1 W, 2 Y Wang, 3 Y Chen. 1 Pediatrics, The Second Affiliated Hospital of Shantou University Medical College, Shantou, China; 2 Pediatrics, Capital Institute of Pediatrics, Beijing, China; 3 Pediatrics, Guangdong Maternity and Child Health Care Hospital, Guangzhou, China

10.1136/archdischild-2014-307384.1017