As Klinefelter's is not commonly associated with severe learning disabilities, they were enrolled in the 100,000 genome project. Whole exome sequencing revealed a mutation involving the SOX5 gene on chromosome 12p12, which is associated with Lamb-Shaffer syndrome. Lamb-Shaffer syndrome is a neurodevelopmental disorder characterised by global developmental delay, learning disability, poor expressive speech and mild dysmorphic facial features (Nesbitt et al 2015). Less than 100 cases have been reported since the condition was first described in 2012.

This new genetic diagnosis has enabled the health care professionals and the parents to understand more about the clinical features, including what surveillance and investigations the patients may require based upon other reported cases with the same mutation.

For parents and patients, having a named diagnosis allows them to connect via support groups with other families who have been diagnosed with the same condition. Historically the 'diagnostic odyssey' in a child with a rare disease could continue for many years, with serial testing of different genes, often with no answers found for patients or their parents. Learning disability is genetically heterogeneous and has previously presented challenges for molecular diagnoses using traditional genetic testing methods. Next generation sequencing has made it possible to sequence millions of fragments of DNA simultaneously, enabling the sequencing of whole patient exomes as part of the 100,000 genome project. These patients are two of many new genetic diagnoses that are a result of the genome project, which are highlighting the utility of exome sequencing in clinically and genetically heterogeneous conditions such as learning disability.

In this unusual case these twins have two different genetic underlying conditions, which demonstrates the importance of investigating further possible causes when previous diagnoses do not explain the evolving clinical phenotype.

G68(P) ABSTRACT WITHDRAWN

G69(P) PARENT-DELIVERED STRATEGIES TO IMPROVE EATING, DRINKING AND SWALLOWING DIFFICULTIES OF YOUNG CHILDREN WITH NEURODISABILITY: CURRENT PRACTICE AND FUTURE RESEARCH

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Aims Eating, drinking and swallowing difficulties (EDSD) are common in young children with neurodisability. EDSD can be caused by physical and non-physical difficulties leading to inadequate calorie intake, affecting a child and parents everyday life. Guidance on how to address these difficulties is

lacking. We undertook a study to answer the question: What interventions, which could be delivered at home by parents, are available to improve eating in young children with neurodisability and are suitable for investigation in pragmatic trials?

Methods We undertook: 1. Updates of three systematic reviews, and a mapping review of the research evidence for interventions and outcome measures. 2. Two national parent and professional surveys: one about current professional practice and what is received by parents, and a Delphi survey to identify the most important outcomes. 3. Focus groups with parents, professionals and young people to discuss current practice and workshops identify future directions and research.

Results 951 parents and professionals participated. The limitations of the research evidence regarding interventions and tools to measure outcomes was identified. The survey showed a wide range of interventions are used in NHS practice – parents and professionals reported variability in provision. Nonetheless, parents and professionals considered many interventions were important, and led to change in EDSD. They identified 19 interventions as important to deliver when needed, and 10 outcomes that were most important to measure (including nutrition, growth, and health/safety); young people agreed these were important outcomes. Parents and professionals were positive about a proposed 'toolkit' of interventions that through shared decision making, could be offered by health professionals.

Conclusions The challenges for future clinical trials of a toolkit of interventions were evident (for example, the range of interventions that children in a comparator group might receive). Parents and professionals thought that implementation of the toolkit as part of usual practice would be a positive step. However, this would require the toolkit to be operationalised through further development – evaluation of its effectiveness could then be undertaken.

G70(P) NITROUS OXIDE ABUSE RESULTING IN SUB-ACUTE DEGENERATION OF THE SPINAL CORD – A CASE REPORT

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A case report of a 15 year old boy presenting with a 3 week history of progressive ataxia and sensory loss. Examination revealed complete bilateral sensory loss from toes to mid-thigh and sensory loss in both little fingers. He admitted to heavy inhalational abuse of nitrous oxide and daily use of cannabis. No other medical problems and no family history of note. Blood tests revealed a normal total vitamin B12 level (247 ng/L), but raised methylmalonic acid (500 nmol/L) and reduced active vitamin B12 (30 pmol/L), this was indicative of an active vitamin B12 deficiency. An MRI demonstrated high signal changes in the posterior cord extending from C1 to the lower thoracic cord which was typical of sub-acute degeneration of the spinal cord. Vitamin B12 deficiency is known to cause sub-acute combined degeneration of the spinal cord. He was treated with daily vitamin B12 injections and showed some clinical recovery whilst an inpatient.