Conclusions Sequential interventions involving staff education and an evidence-based protocol have significantly improved outcomes for patients with polyhydramnios.

433 A PATIENT WITH AUTISTIC SPECTRUM DISORDER AND 17P12 DUPLICATION – LITERATURE REVIEW

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10.1136/bmjpo-2021-RCPCH.244

Background There is an increased awareness in the role of microarray analysis in conditions such as ADHD and autism due to arising association of genetic mutations with these diagnoses as risk factors. We have a patient who has been recently diagnosed with autistic spectrum disorder with duplications in the short arm of chromosome 17 with breakpoints within 17p12.

Objectives The purpose is to use the medical literature to search for other cases of 17p12 duplication to see if there is any phenotypic resemblance between the genetic mutation and neuropsychiatric disorders.

Methods We conducted a literature search for other additional cases of 17p duplication as well as specifically looking for 17p12 duplication. We used database engines such as PubMed, National Centre for Biotechnology Information and the Cochrane Library as well as using information from Unique.

Results 17p12 duplication is a rare disorder with approximately 50 people having been diagnosed in the medical literature. With 17p there are four groups that patients tend to fall into depending on specific breakpoints. With 17p12 can have an association with Charcot-Marie Tooth Type 1a disease if the peripheral myelin protein (PMP22) gene is also included. Duplications with less than 1Mb are strongly associated with autistic spectrum disorder.

Due to small numbers there is very little data and literature regarding 17p duplication in general and even less on 17p12 specifically. Each patient has a unique phenotypic presentation with varying signs and symptoms. This includes behavioural difficulties (such as autistic features and hyperactivity), intellectual disability and unique facial features.

Our patient initially presented with features of learning difficulties, social communication difficulties, sleep disturbances, repetitive behaviours and being investigated for possible dyspraxia. The neonatal period was uneventful and had normal gross motor development. The duplication was approximately 1.3Mb and was stated on the results form as being associated with Charcot-Marie-Tooth Syndrome Type 1a (CMT1A) (CMIM #118220). However, it did not specify as to whether the PMP22 gene was included. CMT1A is a 1.5Mb duplication on 17p12 and our patient's duplication was measured at 1.3Mb. This was thought to be a co-incidental finding for the referral reason of autistic spectrum disorder. There is no family history of Charcot-Marie-Tooth and our patient has no symptoms of peripheral neuropathy.

Conclusions 17p12 duplication is a rare genetic mutation with very little cases within the medical literature. However, as chromosomal microarray has become increasingly requested for patients with unexplained intellectual disability or autistic spectrum disorders there may well be further individuals diagnosed in the future.

434 STREPTOCOCCUS GALLOLYTICUS SUBSPECIES PASTEURIANUS INFECTION IN INFANTS – A SYSTEMATIC REVIEW AND META-ANALYSIS

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10.1136/bmjpo-2021-RCPCH.245

Background *Streptococcus bovis* is a commensal colonic pathogen that is found in both human and animals. *Streptococcus bovis* biotype II/2, which is currently known as *streptococcus gallolyticus* subspecies (ssp) *pasteurianus* (SGP), is associated with sepsis and meningitis in neonates and infants. While this is uncommon, it has been associated with long term morbidity and mortality.

Objectives The aim of this study is to describe the clinical course and outcomes of infants with SGP infection.

Methods This is a systematic review and meta-analysis of studies identified from PubMed/Medline, Embase, Scopus, Web of Science, Google Scholar, and Cochrane Library. The following MeSH keywords were used: 'streptococcus gallolyticus', 'streptococcus bovis', 'newborn', 'infant'. Studies were included if they described infants with culture-proven *streptococcus gallolyticus* (in blood or cerebrospinal fluid) up to 1 year of life. Data extracted included perinatal factors, clinical presentation, investigations, treatment received, and outcomes. Subgroup analyses were performed according to early and late onset of infection, and preterm compared to term infants.

Results A total of 42 articles were identified, which included a total of 97 cases, of which 59 had *streptococcus bovis*, and 38 with SGP infection (i.e. *streptococcus bovis* biotype II/2). Of the 97 cases, they were predominantly term infants (65.7%) male (54.5%), and were born via vaginal delivery (67.4%). The most common clinical symptom at presentation was lethargy (63.3%, 95% CI 45.1%–78.4), fever (60.3%, 48.3–71.2%), irritability (57.6%, 35.1–77.4%), tachypnea (58.1%, 40.4–73.9%) and respiratory distress (48.3%, 21.9–68.0%). There were 28 early-onset infections (≤ 3 days of life), which commonly presented as bacteremia (64.3%). Compared to late-onset infection, neonates with early-onset infection presented with more respiratory symptoms [pooled-risk difference for tachypnea 41.7% (11.2–72.2%), respiratory distress 32.4% (0.5–64.2%)]. The median inflammatory markers at onset of symptoms were not raised - median leukocyte 7000/microL (3490–16100/microL), neutrophils 45% (34–60%), C-reactive protein 9.6mg/dL (5.2–13.48 mg/dL). CSF studies in neonates with meningitis showed predominant pleocytosis (median leukocyte 824/mm³ (13–3016/mm³)). Most cases were susceptible to penicillin (66.1%, 53.6–76.8%) and were treated with a penicillin derivative with good recovery. The reported cases of meningitis had cranial imaging findings of intraventricular hemorrhage, ventriculitis, hydrocephalus, as well as frontal and subdural effusion. There were 4 mortalities - two presented and died on the first day of life, and two had a late-onset presentation and deteriorated before antibiotics could be given.

Conclusions SGP is an important cause of sepsis and meningitis in neonates. Penicillin derivative is an effective medication for SGP with limited reports of resistance and outcomes appear to be favourable.