AN UNUSUAL CASE OF NEONATAL PROLONGED UNCONJUGATED HYPERBILIRUBINEMIA

Aim Our aim is to present the case of an Irish neonate who presented to our Paediatric Department with prolonged hyperbilirubinemia as a consequence of breast milk quality and Gilbert’s Mutation (UGT1A1*28 mutation).

Methods We describe the clinical features, course of illness and haematological findings, management and the challenges of same and in addition the outcome to date in our patient.

Results An Irish male neonate of non-consanguineous union, with gestation of 36 weeks, unremarkable perinatal history, exclusively breast fed, first came to medical attention at third day of life with DCT-negative unconjugated hyperbilirubinemia requiring phototherapy for two days. He represented with increased bilirubin levels again 3 days later and again at two weeks of age, each time necessitating phototherapy. Systemic examination was normal. Thyroid & liver function tests, full blood count, urine culture, urine for cytomegalovirus and toxicology screen were normal. He was fed exclusive expressed breast milk by bottle and was observed to have high volume intake of 275 ml/kg/day, with overfeeding resulting in vomiting. By 5 weeks of age, he had not regained his birthweight and had dropped from the 25–50th to the 0.4th centile. On further work up, his screen for Hereditary Spherocytosis was negative, haptoglobin was low (<0.10 g/L, liver ultrasound scan was normal. His UGT1A1 enzyme levels were sent the results showed that he was heterozygote for UGT1A1*28 mutation indicating carrier status for Gilbert’s Syndrome.

Breastfeeding was discontinued, and his weight and hyperbilirubinemia improved dramatically.

Conclusion Breast-feeding jaundice is traditionally considered as a consequence of inadequate supply, with breast-milk jaundice secondary to increased recirculation of bilirubin due to deconjugating breast-milk enzymes. This case was unusual in that supply was established to be adequate, rather the nutritional quality of the breast-milk was theorised to be suboptimal, secondary to severe social stressors affecting mother. This, in combination with Gilbert’s syndrome status, resulted in moderate prolonged unconjugated hyperbilirubinemia and failure to thrive in this infant.

It may be helpful for Paediatricians to maintain an index of clinical suspicion for the mutation in children with refractory prolonged unconjugated hyperbilirubinemia. We rarely advocate breast-feeding discontinuation, however in this instance it resulted in dramatic clinical improvement of this infant’s condition.

COW’S MILK PROTEIN ALLERGY IN CHILDREN – CLINICAL PRESENTATION, DEMOGRAPHIC DATA AND FAMILY HISTORY IN A STUDY POPULATION

Introduction Food allergy in children is a frequent topic of clinical studies, considering the increasing number of the affected individuals and the diversity of the symptoms these children demonstrate. Cow’s milk protein allergy is the most common food allergy in the paediatric population, with an estimated prevalence of 2–6% in infants.

Cow’s milk protein allergy can induce a variety of clinical symptoms, therefore establishing the correct diagnosis is often difficult. Beside the various clinical presentation, demographic and additional medical data of the research population and their families were also reviewed in this study.

Methods The study was conducted at the Paediatric Gastroenterology Department of the Balassa János County Hospital in Szekszárd, Hungary. The research population (n=47) included children (0–18 years) with symptoms suggesting cow’s milk protein allergy. This component of our research represents the data from questionnaires filled in by the parents. The evaluation of the results was performed with SPSS statistical software.

Results 47 children were included in our study (57.4% male, mean age: 7.36 years, SD: 4.22).
By reviewing the parents’ medical history, 23.4% of mothers and 25.5% of fathers were diagnosed with either an atopic disease or an inflammatory bowel disease; however, examining the siblings of the affected children, 45.7% are reported to have one of these conditions (12 children do not have any siblings).

In the study population, 8.5% were born prematurely and 85.1% were born at term (6.4% did not provide an answer), 46.8% were exclusively breastfed in the first 6 months of life.

Analysing the data about the forms of clinical presentation, 85.1% showed gastrointestinal symptoms, 63.8% demonstrated skin-related problems, 57.4% presented with respiratory manifestation and 44.7% showed behavioural problems.

Conclusion According to our results, a positive family history for atopic diseases and/or inflammatory bowel disease can increase the risk of developing cow’s milk protein allergy; siblings showed a higher prevalence of food allergy when the parents were affected by the aforementioned entities.

The clinical manifestations represent a wide variety of symptoms which frequently overlap, making the diagnostic process often challenging.

We did not observe a significant effect of feeding (breast-feeding or formula feeding), or being premature on the risk of developing cow’s milk protein allergy in children. However, it is important to emphasize the relatively small number of the study population, which is why we plan to continue the research with a larger study group.

All clinicians correctly identified neonatal onset of constipation as a red flag, but only 73.9% and 52.2% were able to identify abdominal distention with vomiting and ribbon stools, respectively, as red flags. 39.1% of respondents stated that they would not routinely investigate idiopathic constipation, whilst up to 56.5% would perform a coeliac screen. 69.6% stated they would never perform a digital rectal examination. 43.5% correctly identified the recommended pathway for treatment and 52.2% correctly identified the referral criteria. Whilst clinicians felt, on average, moderately confident in managing patients with constipation, up to 56.5% did not have specific facilities in their department.

Conclusion While most respondents correctly identified amber and red flags, and were reasonably familiar with management recommendations, the majority of clinicians would investigate constipation – which NICE does not routinely recommend. These findings suggest that further targeted education, as well as specialist resources, may improve clinicians’ knowledge and confidence in managing this condition and also illustrates guideline fatigue.

Introduction Modern epidemiologic data reveal that the prevalence of celiac disease (CD) is approximately 1% in Europe population. Statistics varies considerably in different countries. The latest Russian clinical recommendations ‘Celiac disease in children’ published in 2016, suggest the screening for CD only among children with type 1 diabetes mellitus and autoimmune thyroiditis. However, there are no population-based studies and data about prevalence of CD in average-risk and high-risk groups in Russia.

Aim To determine prevalence of CD in children with abdominal pain.

Methods Tissue transglutaminase antibodies (anti-tTG) of IgA, IgM and IgG classes were detected by quick celiac test «BIO-HIT», based on immunochromatographic method. 500 children with abdominal pain, aged 3 – 17 years, out patients of the second diagnostic center in St.Petersburg were examined using the quick-test. All positive for CD children underwent histological, genetic and serologic tests, as well as in 20 randomly selected children, who didn’t test positive. Antibodies to deamidated gliadin peptides (anti-DGPs) and anti-tTG were investigated by enzyme immunoassay (ELISA). The study patients were genotyped for HLA-DQB1 and DQA1 alleles using real-time polymerase chain reaction.

Results The test detected 6 positive results (5 females with an average age of 12±2.1 years and one 10 years old boy) in group with abdominal pain. CD in all this subjects was diagnosed and confirmed for first time ever. Quick celiac test «BIO-HIT», yield sensitivity 95% and specificity 100%.

Conclusion The prevalence of CD in children with abdominal pain was 1: 83. Quick celiac test is suitable for screening CD in population-based studies, as well as for the diagnosis of this disease in risk groups.