

**P111 SEVERE STEATOHEPATITIS AS A SIGN OF LYSOSOMAL ACID LIPASE DEFICIENCY IN A RUSSIAN INFANT**

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We examined the infant 2 months with severe steatohepatitis. Considering the burdened hereditary history (death of an older child aged 2.5 months from liver cirrhosis), physical examination data (skin and sclera jaundice, severe hepatosplenomegaly), laboratory results (hyperbilirubinemia, high ALT and AST levels, hypertriglyceridemia), instrumental examination (CT scans revealed a liver low density from +4 to +25 Hounsfield Units), the most likely was the presence of a child with a deficiency of lysosomal acid lipase. The activity of lysosomal acid lipase was markedly reduced to 0.006 nmol/spot/h (norm >0.07 nmol/spot/h), which supported the proposed diagnosis. LAL activity was measured in dry blood spots using a fluorometric enzyme assay. All coding exons as well as the adjacent intron regions of the *LIPA* gene were investigated by Sanger sequencing. As a result, we revealed two undescribed earlier small deletions *c.442del* and *c.817\_818del* leading to the frameshifts in exons 05 and 07 of *LIPA* gene, respectively. According to the international OMIM database, mutations in *LIPA* gene are described in patients with a lysosomal acid lipase deficiency, inherited in an autosomal recessive manner. Thus, the cause of marked steatohepatitis in infants may be lysosomal acid lipase deficiency.

**P112 ASSOCIATION OF INTUSSUSCEPTION AND CELIAC DISEASE IN CHILDREN: A CASE REPORT AND LITERATURE REVIEW**

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We present the case of a 22-month-old child, with an acute onset of celiac disease (CD) and recurrent intestinal intussusception.

The child had a marked loss of appetite for 1 month and episodes of food vomiting, almost at every meal, at a distance of about 2 hours, sometimes associated with short-term crying. An abdominal ultrasound has documented a picture compatible with intestinal intussusception (INI), which spontaneously resolved during the ultrasound examination.

Since the criteria of the ESPGHAN guidelines were not met in order to be able to diagnose celiac disease without biopsy (in particular there were no TTG IgA values > 10 times the norm), esophageal-gastro-duodenoscopy was performed. Histological examination confirmed CD. The child was placed on a gluten-free diet with rapid improvement of the clinical conditions and gradual recovery of appetite.

At home wellness in the first days with subsequent appearance of episodes of crying at night, lasting about 5–10 minutes, all at spontaneous resolution. To progressively increase the episodes of crying it was decided to perform

again an ultrasound of the abdomen with evidence of a image compatible again with a picture of INI. Subsequent ultrasound evaluations, performed in the following days, showed the spontaneous resolution of the described picture. In view of the clinical picture, characterized by recurrent INI, it was decided to resume steroid therapy (budesonide, 3 mg/day), waiting for a response to the gluten-free diet.

After 2 weeks of complete well-being with clear improvement of the clinical condition, steroid therapy was gradually discontinued, with no more episodes of possible intermittent intestinal intussusception.

The prevalence values of intestinal infection in CD are equal to 1.6% in adults and ranging from 0.037% to 1.2% in children, in different studies. In a cohort study, a prevalence of about 1% of INI was detected in children with a new diagnosis of MC, before starting the gluten diet. Given that not all children with CD and abdominal pain have undergone abdominal imaging, this estimate refers to clinically significant cases.

The pathogenetic mechanism is not yet fully understood. The course of INI to the diagnosis of CD is generally benign, with spontaneous resolution of the picture with a gluten-free diet. In recurrent intussusception it is important to consider the treatment with steroids before resorting to a more invasive approach, which should be reserved for cases with symptoms of intestinal obstruction.

**P113 DUPLICATION 15Q11-Q13 SYNDROME IN PATIENTS WITH AUTISM SPECTRUM DISORDER: 5 NEW CASES**

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**Background** Duplication 15q11.2 - q13 characterized by autism spectrum disorder (ASD), intellectual disability, motor delays and epilepsy, particularly infantile spasms. Disease frequency in the population is 2–5: 10000, among people with ASD - 1:250–1:500. The duplication described in as an interstitial duplication 20% cases and as supernumerary isocentric chromosome in 80% of cases.

**Patients and methods** We analyzed 5 patients 15q11-q13 duplication aged 3 to 5 years - 3 girls and 2 boys. We researched the clinical picture in 5 children with 15q11-q13 duplication syndrome. Age of children ranged from 3 years (36 months) averaging 4 years (48.2 months), 3 girls and 2 boys. Array CGH was proved in 4 children, and FISH diagnostic - in one case of supernumerary isocentric chromosome 15.

**Results** We determined the duplication size from 437009 to 4914267 kb. One patients revealed two independent duplication the analyzed area - 15q11.2 (25199239\_25199239) x3, 15q12 (27183835\_27604796) x3. In one case identified variant - ish inv dup (15) (q11.2q13)(D15Z1++, D15S10++).

All patients have development delayed and mental retardation, mild to severe. Speech disorders was varying degrees, one child had echolalia. Behavioural problems were presented with autistic features (3 children), stereotypes (4 children), hyperactivity (3 children), aggression (1 child).

Elements of abdominal obesity noted in one patient. Facial features was represented flat nose, epicanthic folds, broad nose tip, long filtrum.

EEG was performed in 3 patients. Typical epileptiform activity was not registered. Two children had cardiac arrhythmias: recurrent ventricular tachycardia in one patient, recurrent ectopic atrial tachycardia, transient AV block- in another. **Conclusion** Duplication 15q11.2 - q13 was diagnosed in 5 patients with behavioral problem. Two patients have cardiac arrhythmias.

**P114 RECURRENT A&E ATTENDANCES? REMEMBER THE PSYCHOSOCIAL HISTORY**

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In cases of recurrent hospital attendances it is important to stop and think. In cases of recurrent diabetic ketoacidosis it can be a matter of life and death as each subsequent episode is associated with a higher mortality. It is easy to be focused on managing the acute medical emergency but unless we broaden our horizon and consider the psychosocial aspects of our patients we will miss important risk factors for comorbid mental ill health. Here we present a case of diabulimia where the recognition of an eating disorder in the context of type 1 diabetes was the essential step in stopping a recurrent pattern of life threatening diabetic ketoacidosis.

**P115 OVARIAN TORSION IN CHILDREN: CASE REPORT AND DISCUSSION**

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Ovarian torsion is an uncommon cause of acute abdominal pain in the paediatric population and extremely rare in early childhood. The presentation can be non-specific, mimicking other pathologies such as acute appendicitis, resulting in diagnostic delay. The aetiology, diagnosis and management can differ from those in adults. Misdiagnosis of adnexal torsion is not uncommon in children and may cause loss of the ovary or fallopian tube. In the case of acute abdominal pain, ultrasonography is usually the most routinely used diagnostic tool, alongside colour-Doppler for evaluation of blood circulation in the ovarian pedicle.

With the aid of a case report we will discuss the common presentation and examination findings of adnexal torsion along with the causes and risk factors. We will illustrate the normal adnexal anatomy and show the common imaging findings of ovarian torsion. Further imaging and management options will also be reviewed.

The literature has reported up to an 80–90% rate of ovarian salvage with early intervention. Early and accurate diagnosis of ovarian torsion and early intervention with restoration

in blood flow is imperative in avoiding irreversible damage to the ovary and long term fertility issues.

**P116 EXPECTING THE UNEXPECTED: AN EPISODE OF PARADOXICAL SINUS BRADYCARDIA SECONDARY TO ATROPINE ADMINISTRATION IN A PRE-TERM NEONATE**

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**Background** We present the case of a preterm neonate who received two separate doses of Atropine (20 micrograms/kg, no minimum dosing) with subsequent unexpected episodes of bradycardia immediately post administration.

**Case presentation** A 34+1 male triplet, birth weight 1.8 kg, required intubation for surfactant administration at approximately 16 hours of life. The infant was being treated for RDS and early onset sepsis. No episodes of apnoea or bradycardia had been noted. As per local policy, doses of Atropine (20 micrograms/kg), Fentanyl (3 micrograms/kg) and Suxamethonium (2 mg/kg) were prepared and independently checked by an experienced neonatal nurse. Before anaesthesia, IV Atropine was administered via a peripheral IV line. Within 5 seconds of administration, the infant became bradycardic to a rate of 50bpm which was confirmed on auscultation. The infant also appeared apnoeic, unresponsive and mottled, requiring bag-mask ventilation until heart rate recovery at approximately 30 seconds. Intubation attempt was abandoned and the Consultant Neonatologist on-call was asked to attend. Once in attendance, the Consultant prepared for intubation and a further dose of IV Atropine (20 micrograms/kg) was administered. Again immediate bradycardia to <60bpm was observed. Intubation was attempted without use of anaesthetic drugs but was unsuccessful. The infant was subsequently intubated with Fentanyl and Suxamethonium, both having their expected sedative and paralysing effects. Surfactant was administered and the infant was managed on conventional ventilation until extubation the following morning.

**Discussion** Bradycardia is a relatively common occurrence during paediatric intubation. This is usually secondary to hypoxaemia, vagal stimulation during laryngoscopy or the pharmacologic effect of anaesthetic drugs. There is much debate regarding the use of prophylactic Atropine during the pre-medication routine to reduce the incidence of bradycardia or collapse. The evidence for its use is largely observational and somewhat conflicting. The American Heart Association's 2015 PALS Guideline update no longer supports the routine use of Atropine in the pre-intubation of critically ill infants though it may be considered as pre-medication in infants where there is a higher risk of bradycardia. Furthermore, if Atropine is being used, the previous recommendation of a minimum dose of 100 micrograms (largely based on the findings from Dauchet et al) to avoid paradoxical bradycardia is no longer observed. A dose of 20 micrograms/kg, as given to our patient, may be considered.

This case report illustrates the potential complication of paradoxical sinus bradycardia with low dose Atropine. European paediatricians should bear this unexpected complication in mind when deciding to administer this drug.