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

Andrey Surkov*, Anton Anushenko, Goar Movsisyan, Aleksandr Potapov, Elena Kulebina, Kirill Savostyanov, Aleksandr Pushkov, Natal'ya Zhurkova, Nikolaj Mayanskij, Ekaterina Brzhozovskaya. *National Medical Research Center for Children's Health, Moscow, Russian Federation*

10.1136/archdischild-2019-epa.466

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

Martina Mainetti*, Vanna Graziani, Caterina Radice, Federico Marchetti. *Department of Pediatrics, S. Maria delle Croci Hospital, Ravenna, Italy*

10.1136/archdischild-2019-epa.467

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

¹OB Kondakova, ¹DI Grebenkin, ²NV Zhurkova, ¹TT Batysheva, ³N Vashakmadze*. ¹Scientific and Practical Center of Pediatric Psychoneurology Department of Health of Moscow, Moscow, Russian Federation; ²National Medical Research Center of Children's Health of the Ministry of Health of the Russian Federation, Moscow, Russian Federation; ³Pirogov Russian National Research Medical University, Moscow, Russian Federation

10.1136/archdischild-2019-epa.468

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% of 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 duplications 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).