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Effectiveness of surgical treatment in patient with PFAPA and congenital syndrome

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The pathogenesis of the pediatric disorder periodic fever, aphthous stomatitis, pharyngitis and cervical adenitis (PFAPA) syndrome is unknown. It is regarded as an autoinflammatory process. Disease onset is usually before the age of five and generally resolves before puberty with no consequences for the patient. Children are asymptomatic between episodes and show normal growth. No specific diagnostic test for PFAPA is currently available. Syndrome has overlapping symptoms with other periodic fever syndromes with a known genetic cause. Genomic analysis of familial cases by genome-wide linkage analysis and whole-exome sequencing did not reveal rare variants in a single, common gene. In addition, genetic variants that are known to cause other autoinflammatory syndromes have been found in PFAPA patients, but the impact of these genetic variants in PFAPA syndrome is still unknown. In 2-year Caucasian/Azerbaijan girl demonstrated repeated fever episodes with high levels (90-200mg/L) of C-reactive protein (CRP) since 6 months. She was observed regularly because of microcephalus, slight developmental delay and growth retardation, muscle hypotonus and dysmorphic phenotype (broad forehead, hypertelorism, micrognathia and retrognathia, fluffy eyebrows, long and tight eyelashes, long filtrum, narrow lips). On genetic consultation, she was diagnosed with 7p22 microdeletions. During a period of January-October 2018, she was hospitalized 6 times with high fever, cervical/adenitis and sore throat (3 times with aphthous pharyngitis). Different laboratory tests and instrumental investigations were performed and were normal: abdomen ultrasound, chest X-ray, EKG and EHHOKG, ANA, HIV, Borreliosis serology and Quantiferron test, urine test and urine culture. Cervical ultrasound revealed increased lymphoid nodules with normal structure. In a period of January-July of 2018, she received 4 antibiotic courses because of high CRP levels and pharyngitis.

A blood test revealed no neutropenia, sedimentation rate was always increased up to 20-40mm/t, procalcitonin level and blood culture repeatedly negatives. Brain MRI with spectroscopy was performed to exclude intracranial pathology because of congenital problems. ENT repeated consultations excluded otitis media, but adenoid hypertrophy was considered. PFAPA was suspected because of typical clinical symptoms (repeated episodes of fever with aphthous pharyngitis, cervical/adenitis and high CRP levels, absence of neutropenia). Prednisolone treatment 1mg/kg per os was used twice with excellent effect. Adenotomy with tonsillectomy was performed in October 2018. After this treatment in a period of November 2018- May 2019 the patient was ill 4 times with no high fever (gastroenteritis, conjunctivitis, rhinopharyngitis and varicella with otitis media) and just once needed antibiotic treatment. Sequencing of genes was performed to exclude MEFV, MVK, TNFRSF1A, IL1RN and other gene abnormalities, using Illumina TruSight One expanded panel (6700 genes). No monogenic fever syndrome was revealed.

Recent Publications

1. Artyushenko NK, Influence of connective tissue dysplasia on hemodynamics in a maxillary artery and mucosal vessels in children with anomalies and deformations of maxillofacial area.
2. Artyushenko, Antonova (2011) New technologies in stomatology, International conference of maxillofacial surgeons and stomatologists. Page 28-29.
3. Antonova N.S (2011) Features of clinical manifestations of CTD in children with congenital anomalies and deformations of maxillofacial area/N.S. Antonova/Materials XVI of the International conference of maxillofacial surgeons and stomatologists "New technologies in stomatology" Page 26-27.
4. Antonova N.S (2012) Features of treatment of children with congenital anomalies and acquired deformations of maxillofacial area and accompanying connective tissue dysplasia, Page 86-87.

Biography

Natalia Antonova completed her PhD in the year 2013 from North-Western State Medical University named after I.I. Mechnikov, St-Petersburg, Russia. She works as a Paediatrician and children at the Tallinn Children's Hospital. She is an author of 15 articles and 10 oral presentations and lectures for family doctors in Estonia. In 2011 she completed clinical attachment in paediatric at Al Wasl Maternity and Paediatric Hospital in Dubai, UAE. She is a member of Open American/ Austrian University and participated in 2 in Saltzburg CHOP seminar in Pediatric Pulmonology in 2010 and Pediatric Infectious Diseases in 2017 with the clinical cases presentations. X.2016-VI.2017 she completed a EULAR on-line course in Paediatric Rheumatology.

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