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World Pediatrics 2019: Effectiveness of surgical treatment in patient with PFAPA and congenital syndrome - Natalia Antonova - Tallinn Children´s Hospital

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Introduction: The pathogenesis of the pediatric disorder periodic fever, aphthous stomatitis, pharyngitis, cervical adenitis (PFAPA) syndrome is unknown. It is regarded as an auto inflammatory process. 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 auto inflammatory syndromes have been found in PFAPA patients, but the impact of these genetic variants in PFAPA syndrome is still unknown.

A 2-year Caucasian/Azerbaijan girl demonstrated repeated fever episodes with high levels (90-200 mg/l) of C-reactive protein (CRP) since 6mo. 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 l/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 20-40 mm/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. A 2-year Caucasian/Azerbaijan young lady showed rehashed fever scenes with significant levels (90-200 mg/l) of C-receptive protein (CRP) since 6 mo. She was noticed consistently on account of microcephalus; slight formative postponement and development hindrance, muscle hypotonus and dysmorphic aggregate (wide brow, hypertelorism, micrognathia, and retrognathia, fleecy eyebrows, long and tight eyelashes, long filtrum, slender lips). On hereditary meeting, she was determined to have 7p22 microdeletions. During a time of January-October 2018, she was hospitalized multiple times with high fever, cervical l/adenitis and sore throat (multiple times with aphthous pharyngitis). Diverse lab tests and instrumental examinations were performed and were typical: midsection ultrasound, chest X-beam, EKG and EHHOKG, ANA, HIV, Borreliosis serology and Quantiferron test, pee test, and pee culture. Cervical ultrasound uncovered expanded lymphoid knobs with typical structure. In a time of January-July of 2018, she got 4 anti-infection courses on account of high CRP levels and pharyngitis. A blood test uncovered no neutropenia, Sedimentation rate was constantly expanded up to 20-40 mm/t, procalcitonin level and blood culture consistently negatives. Cerebrum MRI with spectroscopy was performed to bar intracranial pathology as a result of innate issues. ENT rehashed meetings barred otitis media, yet adenoid hypertrophy was thought of. Prednisolone treatment 1 mg/kg per os was utilized twice with superb impact. PFAPA was associated on the grounds that with run of the mill clinical side effects (rehashed scenes of fever with aphthous pharyngitis, cervical l/adenitis and high CRP levels, nonappearance of neutropenia).

Prednisolone treatment 1 mg/kg per os was utilized twice with superb impact. Adenotomy with tonsillectomy was acted in October 2018. After this treatment in a time of November 2018-May 2019 the patient was sick multiple times with no high fever (gastroenteritis, conjunctivitis, rhinopharyngitis and Varicella with otitis media) and just once required anti-infection therapy. Sequencing of qualities was performed to avoid MEFV, MVK, TNFRSF1A, IL1RN, and other quality anomalies, utilizing Illumina TruSightOne extended board (6700 qualities). No monogenic fever condition was uncovered. PFAPA was suspected because of typical clinical symptoms (repeated episodes of fever with aphthous pharyngitis, cervical l/adenitis and high CRP levels, absence of neutropenia). Prednisolone treatment 1 mg/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 also other gene abnormalities, using Illumina TruSightOne expanded panel (6700 genes). No monogenic fever syndrome was revealed.

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