FRI0578

LACC1 MUTATION IN THREE SIBLINGS WITH POLYARTHRITIS WITHOUT SYSTEMIC MANIFESTATIONS

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Background: Juvenile idiopathic arthritis (JIA) is a complex group of disorders characterized by wide phenotypic diversity and genetic heterogeneity. It has multifactorial etiology varying among different subtypes. There are emerging reports on new gene loci being identified especially in families with many affected members (1,2)

Objectives: To report 3 siblings with polyarthritis who were detected to have mutation in Laccase domain containing 1 (LACC1) gene.

Methods: We reviewed case records of 3 siblings with infantile onset polyarthritis and performed whole exome sequencing on their DNA samples for LACC1, MMP2 and WISP genes.

Results: A non-consanguineous family from north-western part of India reported with 3 daughters having polyarticular joint disease. The chronology of symptoms was similar in all 3 children. Joint symptoms started in later half of infancy with swelling of knee and ankle. This disease was progressive and over the following 2 years there was involvement of small joints and cervical spine too (Figure 1) along with deformities and contractures. The investigations at the time of first evaluation are given in Table 1. Radiographs in all 3 sisters showed osteoporosis and erosion of vertebrae (Figure 2).

The children were initially started on naproxen. Later, in view of rheumatoid factor positivity in eldest sister we decided to give them a trial of oral prednisolone (initially 1 mg/kg and subsequently tapered) and subcutaneous methotrexate (10 mg/m²/week). Methotrexate was continued on long term basis. Over the next 24 months of follow-up the clinical improvement has been sustained.

Whole-exome-sequencing in all three girls has revealed mutation in LACC1 gene [Exon 4; c.832G>C; p.(Ala278Pro), Homozygous, Autosomal recessive].

Conclusion: This case report further provides evidence to the emerging association of LACC1 mutation with familial aggregation of JIA (1,2). Long term follow-up of these patients is necessary to determine the natural course of this intriguing clinical entity.

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Disclosure of Interests: None declared

DOI: 10.1136/annrheumdis-2019-eular.6587

FRI0579

PREDNISONE VERSUS STANDARD THERAPY IN SYDENHAM'S CHOREA: RESULTS FROM A RETROSPECTIVE STUDY

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Background: Sydenham's chorea is a major and delayed manifestation of acute rheumatic fever [1] and is considered to be a prototype of an autoimmune disorder triggered by an infectious agent. Aside from conventional symptomatic treatment (carbamazepine, valproate, neuroleptics), the use of steroids has also been advocated, mainly in severe, drug-resistant cases or if clinically disabling side effects develop with first line therapies. However the evidence of corticosteroids efficacy is weak.

Objectives: To describe the efficacy of corticosteroids therapy versus standard therapy in children affected by Sydenham's Chorea in a cohort of Italian patients with acute rheumatic fever (ARF).

Methods: A retrospective observational study was conducted. The hospital records for all children diagnosed as having ARF between January 2007 and December 2017 in the Pediatric Rheumatology Division of the Meyer Children Hospital (Florence) and IRCCS Burlo Garofolo (Trieste) were reviewed. The diagnosis of ARF was made by pediatric rheumatologists and all the children discharged with a ICD 9 code consistent with ARF were included. For the purpose of the study only patients with Sydenham's Chorea were enrolled. Files were analyzed for demographic data, clinical and laboratory findings on admission and during the hospital stay and patients with incomplete information were excluded. Time and resolution of choreic symptoms were evaluated in consideration of presence or absence of corticosteroid therapy.

Results: Thirty patients were enrolled; 23 out of 30 (76%) had generalized chorea. 15 (50%) were treated with prednisone (2 mg/kg/day in a single administration per day for 14 days before tapering), 11 (36.6%) were not treated with medications and 4 (13.3%) received pimozide or sodium valproate. We considered together patients who did not receive any specific therapy for chorea and patients who received only symptomatic anti-chorea drugs (pimozide in 3 cases and sodium valproate in 1 case). Therefore we obtained two groups of 15 participants each: "Prednisone group" and "Standard therapy group". The time required for clinical improvement in the two groups appeared statistically different (P = 0.002). In "Prednisone group" the median time for improvement was 4.0 days (interquartile range: 3) whereas in "Standard therapy group" it was 16.0 days (interquartile range: 16). Furthermore, the "Prednisone group" had a median remission time of 30.0 days (interquartile range: 34) whereas "Standard therapy group" presented a median remission time of 135.0 days (interquartile range: 309) (P < 0.001). At least one episode of relapse occurred in 1 (6.7%) out of 15 patients in the "Prednisone group" and in 3 patients (20%) in the "Standard therapy group". However, the difference is not statistically significant (p = 0.598).

Conclusion: Our study shows that corticosteroid therapy is associated with a faster resolution of Sydenham's Chorea's symptoms compared to treatments considered of first line, such as: no therapy, valproate or pimozide.

Disclosure of Interests: Elena Favaretto: None declared, Giulia Gortani: None declared, Gabriele Simonini Grant/research support from: Abbvie, Speakers bureau: Abbvie, Serena Pastore: None declared, Rolando Cimaz: None declared, Alberto Tommasini: None declared, Andrea Taddio: None declared

DOI: 10.1136/annrheumdis-2019-eular.8181

FRI0580

ULTRASONOGRAPHY SCORE CORRELATES WITH DISEASE ACTIVITY IN JUVENILE IDIOPATHIC ARTHRITIS – ENTHESITIS RELATED ARTHRITIS – A CROSS SECTIONAL OBSERVATIONAL STUDY FROM INDIA

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Background: Enthesitis is a distinct clinical hallmark of spondyloarthropathy in children and adults. Ultrasonography (US) is a powerful tool to detect enthesitis. There are only a few studies on US detected enthesitis in Juvenile Idiopathic Arthritis - Enthesitis Related Arthritis (JIA-ERA) from India.

Objectives: To assess the presence of clinical and US detected enthesitis in JIA- ERA and to correlate with disease activity.

Methods: 51 consecutive JIA-ERA patients fulfilling the ILAR classification criteria were evaluated clinically and sonographically from October 2016 to September 2017 in a tertiary care centre from Northern India. Enthesitis was clinically assessed using the MASES (Maastricht Ankylosing Spondylitis Enthesitis Score). Additionally peripheral enthesal sites of lower limb (insertion of plantar fascia at calcaneal tuberosity, insertion of Achilles at superior pole of calcaneus, quadriceps tendon at superior pole of patella and patellar tendons proximally at inferior pole of patella and distally at anterior tibial tuberosity) were clinically and sonographically assessed for enthesitis. The clinical assessment included JADAS 27 and BASDAI for disease activity and BASFI for function. Ultrasound was performed with a multi-frequency linear-array transducer (8-13MHz) using B-mode settings of dynamic range (40-50dB); GS- frequency (11-13 MHz); GS gain (60 dB). Power Doppler (PD) mode settings used: pulsed repetition frequency (400-800 Hz); PD-gain (highest possible gain with minimum background noise). US parameters (hypo echogenicity, increased thickness of entheses, intratendinous calcification, enthesophyte and erosion(s)) were assessed according to OMERACT 2014 definitions of ultrasonography