REFERENCES:

- [1] Vojinovic J, Cimaz R. Pediatric Vitamin D update for the pediatric rheumatologists. *Rheumatol* 2015;13:18.
- [2] Stagi S, Cavalli L, Bertini F, et al. Vitamin D levels in children, adolescents, and young adults with juvenile-onset systemic lupus erythematosus: a cross-sectional study *Lupus* 2014; 23:1059–1065.

Disclosure of Interest: None declared

DOI: 10.1136/annrheumdis-2018-eular.1070

THU0592

PUBERTAL DELAY DESPITE INTENSIVE TREATMENT OF JUVENILE IDIOPATHIC ARTHRITIS: RESULTS OF A LONGITUDINAL STUDY

P. Van Pelt^{1,2}, A. Hokken-Koelega³, R. Dolhain⁴, A. Kruize⁵, H. Bijlsma⁵, N. Wulfrat². ¹Rheumatology and paediatric rheumatology, Erasmus Medical Centre Rotterdam, Rotterdam; ²Paediatric immunology and rheumatology, University Medical Centre Utrecht, Utrecht; ³Paediatric endocrinology; ⁴Rheumatology, Erasmus Medical Centre Rotterdam, Rotterdam; ⁵Rheumatology, University Medical Centre Utrecht, Utrecht, Netherlands

Background: Delayed puberty and reduced adult height have been reported in JIA before the era of biologicals. Long-term consequences of delayed puberty are among others growth disturbances, low bone mineral mass and decreased fertility. Treatment with anti-TNF restores growth, but data on puberty are unknown

Objectives: We evaluated onset and course of puberty and growth, in JIA-patients who are treated intensively, including the possibility of biologicals, and identified variables related with puberty and growth

Methods: In a longitudinal JIA-cohort, all consecutive patients (10–21 years) were followed for three years. Annual examinations were performed regarding demographic and disease-related items as well as Tanner pubertal stages and anthropometric measurements. Median ages at reaching each stage were estimated by Kaplan-Meier curves. Parametric tests were used to determine differences between patients and healthy controls, non-parametric tests between patient-groups. Univariate analyses and mixed models were used to identify associated variables

Results: 138 patients were included (66% girls). Median disease-duration was 7.8 years (IQR 3.7–10.5), median JADAS-27 3.7 (IQR 1.3–8.0), DAS-28 2.16.^{1,5–2,8} Puberty onset was 1.2 years delayed in girls ($p<0.01$), in boys 0.6 years (ns). The progression was also delayed: end-stage (Tanner-5) in girls was 3.3 years delayed ($p<0.01$), in boys 1.7 years ($p<0.01$). A positive association was found for longer disease-duration and a lower BMI. Biological-use was not associated. Both for girls and boys, standardised height was not different at the onset and at the end of puberty

Abstract THU0592 – Figure 1 Vertical. lines indicate median age of healthy population of Tanner stages

Conclusions: In contrast to normalised longitudinal growth, we found in JIA-patients in disease remission or with low disease activity a delayed onset and progression of puberty despite intensive treatment including biologicals. Eventually, puberty was completed and normal adult height was reached. The effects on bone mass and fertility will have to be evaluated in cohort studies

Acknowledgements : This study is financially supported by the Dutch Arthritis Association

Statistical support was provided by Reinier Timman and Kees A.G.M.van Montfort, department of biostatistics, Erasmus University Rotterdam.

Disclosure of Interest: None declared

DOI: 10.1136/annrheumdis-2018-eular.4227

THU0593

MULTICENTRIC OSTEOLYSIS WITH NODULOSIS AND ARTHROPATHY (MONA): REPORT OF THE FIRST LEBANESE FAMILY

R. Dagher¹, E. Saliba², M. Rizkallah², M.C. Fadous Khalife¹, A. Megarbane³. ¹pediatrics, Notre Dame de Secours University Hospital, Byblos; ²orthopedics, Hotel Dieu de France University Hospital, Beirut, Lebanon; ³medical genetics, Institut Jérôme Lejeune, Paris, France

Background: “Multicentric Osteolysis with Nodulosis and Arthropathy” (MONA) also known as Winchester-Torg syndrome is a rare chronic skeleton disorder caused by matrix metalloproteinase 2 (MMP2) deficiency. It is characterised by facial dysmorphism, subcutaneous fibrocollagenous nodules, carpal and tarsal osteolysis and interphalangeal joint erosions. Short stature and Osteopenia are frequent and heart defects have been described. As children first present with joint pain, swelling and stiffness, MONA is often misdiagnosed as Juvenile Idiopathic Arthritis (JIA).

Objectives: We report the first Lebanese family with 3 siblings presenting MONA, two of which were diagnosed at first as JIA.

Methods: The proband is the eldest boy born of consanguineous parents. At birth, a ventricular septal defect (VSD) was noted. At the age of 5 bone erosions and nodules in his hands and feet, and cuneiform vertebrae of unknown cause appeared. He was diagnosed with JIA. His pain partially improved with methotrexate and TNF agonist treatment; steroid injections were performed in wrists and permitted a gain in range of motion. Secondly, Osteopenia was detected.

The middle boy was examined at the age of 7; he had wrist arthritis and metacarpal tenosynovitis. He was known to have psoriasis. He was treated first with steroid injections then with NSAID and methotrexate with a good response but his condition slowly progressed to deviated fingers.

The youngest boy suffered from foot pain and Kohler disease was diagnosed at the age of 4. Soon after he developed global stiffness of the foot with erosions and nodules. He had failure to thrive and a VSD was detected.

Results: This family history along with the progressive coarsening of face features in the 3 siblings raised the possibility of a genetic disorder. Exome sequencing for skeletal dysplasia in the eldest boy detected a mutation in the MMP2 gene (NM_001127891:exon2:c.8A>G:p.Y3C). The same mutation was found in the 2 other siblings.

Conclusions: These cases are to add to the 44 individuals coming from 27 different families, with molecularly proven MONA reported in the medical literature. This diagnosis should be raised in front of patients having resistant articular erosions with hand and feet nodules, despite anti-rheumatic drugs. Till date, no specific therapy is available and management is only supportive. In this described family, steroid injections were efficient when children presented with swelling and stiffness of joints and seemed to slow progression to erosion.

REFERENCES:

- [1] Bhavani GS, Shah H, Shukla A, Gupta N, Gowrishankar K, Rao AP, et al. Clinical and mutation profile of multicentric osteolysis nodulosis and arthropathy. *Am J Med Genet A* 2016 Feb;170A(2):410–7.