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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

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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.

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THU0594

SHIARI-JAVADI CRITERIA FOR THE DIAGNOSIS OF GENERALISED JOINT HYPERMOBILITY IN CHILDREN

R. Shiari, V.J. Parvaneh. *Pediatric Rheumatology, Shahid Beheshti University of Medical Sciences, Tehran, Iran, Islamic Republic of Iran*

Background: Benign joint hypermobility syndrome (BJHS) is the occurrence of musculoskeletal symptoms in hypermobile individuals in the absence of systemic rheumatologic disease.^{1–3} One of the most common criteria for the evaluation of generalised joint hypermobility is Beighton's criteria. However, Beighton criteria were designed for all ages not specifically for children group (according to the children physiological and growth characteristics). Inability to identify limited hypermobility, failure to differentiate generalised joint hypermobility from joint hypermobility syndrome and BJHS and the restrictions on the use of epidemiological studies are further limitations.^{4, 5}

Objectives: Because of the disadvantages of the existing criteria and the high prevalence of generalised joint hypermobility in children, we decided to propose further modifications to Beighton criteria for the diagnosis of joint hypermobility.

Methods: A case-control study was designed with 200 participants from 3 to 16 years of age with 100 children with BJH (according to Beighton criteria) in case group and 100 age-sex matched children as control group. Cases were selected from outpatients Clinic of Rheumatology and the control group was selected from the emergency department of hospital. The case group consisted of children who had musculoskeletal pain complaints or were suspected to be hypermobile. The Beighton criteria were used as the gold standard, and all of cases fulfilled the Beighton criteria. In addition, the participants in the control group should be free of chronic disease or musculoskeletal complaints. For the determination of new criteria we performed literature review on the range of motion in different age and gender paediatric groups, studied the various existing criteria and their differences, and utilised our own clinical observations. Eight manoeuvres were set for examination (Fig 1). All were bilateral, except the neck hyperextension.

Results: There were 42 (42%) male children in each group. The mean age was 6.8 years. Table 2 compares the results of the new and Beighton criteria in both cases and controls. All cases were hypermobile, and two of the 100 controls were hypermobile with the new criteria. Based on these results, new proposed criteria had the sensitivity, specificity, positive predictive value and negative predictive value of 100%, 98%, 100% and 98% respectively.

Conclusions: Shiari-Javadi criteria appears to be useful for detecting hypermobility. In addition, they have also overcome many of the disadvantages of the Beighton criteria, and are easier, more practical and more comprehensive to be used in children. However, further studies are required to validate this criteria.

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THU0595

CLINICAL AND THERAPEUTICAL DIFFERENCES BETWEEN ADULTS AND CHILDREN IN A COLOMBIAN COHORT WITH INFLAMMATORY IDIOPATHIC MYOPATHY

R. Fuentes¹, M. Cañola¹, J.C. Diaz-Coronado², A. Rojas-Villarraga², D. Hernandez-Parra², P. Perez-Estrada², J.C. Salazar-Urbe³, R. Pineda-Tamayo².
¹Rheumatology Department, ²Clinical Research, Artmédica, ³National University of Colombia, Medellín, Colombia

Background: Age-related autoimmunity is a key differentiating factor with diverse presentations in different age groups possibly depending on immune regulatory mechanisms. Even patients with the same diagnostic category may have different presentations according to age and other factors. Inflammatory Idiopathic Myopathies (IIM) are not the exception with important clinical and prognostic differences between adults and children.

Objectives: To describe the clinical and therapeutic differences between adults and children (younger than 18 years old) in a cohort with IIM

Methods: A cross-section retrospective research was done with data collected between 2014 and 2017 from a population diagnosed with IIM according to Peter and Bohan criteria and followed up for at least six months. Statistical association was evaluated by means of Chi-square and Fisher's tests.

Results: Out of a total of 149 patients, 112 were adults and 37 were children. There was significant difference between the clinical presentation of symmetrical muscle weakness 98% in adults vs 73% in children (p: 0.0001), Gottron's papules 32% vs 89% (p: 0.0001), Heliotrope rash 31% vs 62% (p: 0.0016), Calcinosis cutis 1% vs 37% (p: 0.0001), Dermatological involvement 47% vs 94% (p: 0.0001), Myopathic changes in the electromyogram 41% vs 25% (p: 0.0001), and Myopathy proven biopsy 41% vs 25% (p: 0.0001). Therapeutic options showed a significant difference in the use of steroid therapy 93% vs. 84% (p: 0.045), Methotrexate (58% vs 91% p: 0.0001), Azathioprine (62% vs 27% p: 0.0002) and Chloroquine (31% vs 73% p: 0.0001). Other therapies like Cyclophosphamide (19% vs 5% p: 0.04) and Immunoglobulins (23% vs 5% p: 0.01) were significantly different between adults and children.

Abstract THU0595 – Table 1. Clinical characteristics of Colombian Patients with Inflammatory Idiopathic Myopathy (Adults and Children)

	Juvenile n=37		Adults n=112		p-value
	N	%	N	%	
Female	28	75,7	76	67,9	0,42
Clinical characteristics					
Symmetrical muscle weakness	27	73	110	98,2	<,0001
Gottron's papules	33	89,2	49	32,9	<,0001
Heliotrope rash	23	62,2	35	31,3	0,0016
Calcinosis cutis	14	37,8	2	1,8	<,0001
Dermatological involvement	35	94,6	53	47,3	<,0001
Myopathic changes (EMG)	9/23	39,1	68/81	84,0	<,0001
Biopsy-proven myopathy (Positive)	4/16	25	46/54	41,1	<,0001
ANA(+)	14/27	51,9	65/89	73,0	0,0580

EMG, electromyogram

Conclusions: Several differences in the clinical and therapeutic characteristics were found between adults and children with IIM. Adults had more frequently symmetrical weakness, myopathic changes in EMG and biopsy proven myopathy. In contrast children had higher skin involvement. Calcinosis constituted an important manifestation of IIM in children. These results suggest that paediatric IIM is a distinct sub-phenotype related to the early age at onset and possibly mediated by different immune interplay key factors.