REFERENCES:


Disclosure of Interest: None declared


THU0592

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

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Background: Delayed puberty and reduced adult height have been reported in JIA before the era of biologics. 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.

Objective: We evaluated onset and course of puberty and growth, in JIA-patients who are treated intensively, including the possibility of biologics, 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 (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

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 biologics. 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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Disclosure of Interest: None declared


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

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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).

Objective: 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 ventriculoseptal 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. Secondarily, 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:

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

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Background: Benign joint hypermobility syndrome (BJHS) is the occurrence of musculoskeletal symptoms in hypermobile individuals in the absence of systemic rheumatologic disease. 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 children physiological and growth characteristics). Inability to identify limited hypermobility in children and the lack of studies of the various existing criteria and their differences necessitated the current study.

Methods: A case-control study was designed with 200 participants from 3 to 16 years of age with 100 children with BJHS (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 rheumatologic disease.

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.

REFERENCES:

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

<table>
<thead>
<tr>
<th></th>
<th>Juvenile n=37</th>
<th>Adults n=112</th>
<th>p-value</th>
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<tbody>
<tr>
<td>Female</td>
<td>28</td>
<td>75,7</td>
<td>67,9</td>
</tr>
<tr>
<td>Clinical characteristics</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Symmetrical muscle weakness</td>
<td>27</td>
<td>73,110</td>
<td>98,2</td>
</tr>
<tr>
<td>Gottron’s papules</td>
<td>33</td>
<td>89,49</td>
<td>32,9</td>
</tr>
<tr>
<td>Heliotrope rash</td>
<td>23</td>
<td>62,35</td>
<td>31,3</td>
</tr>
<tr>
<td>Calcinosis cutis</td>
<td>14</td>
<td>37,82</td>
<td>2,1</td>
</tr>
<tr>
<td>Dermatological involvement</td>
<td>35</td>
<td>94,53</td>
<td>47,3</td>
</tr>
<tr>
<td>Myopathic changes (EMG)</td>
<td>9/23</td>
<td>39,68</td>
<td>84,0</td>
</tr>
<tr>
<td>Biopsy-proven myopathy (Protein)</td>
<td>4/6</td>
<td>25,46</td>
<td>41,1</td>
</tr>
<tr>
<td>ANA(+)</td>
<td>14/27</td>
<td>51,965</td>
<td>73,0</td>
</tr>
</tbody>
</table>

EMG, electroneurogram

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. Calcinosus 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.