Idiopathic multicentric carpalosternal osteolysis (MCTO) is a congenital disease, likened to MAFB-gene mutation, described in 2012. MCTO is characterized by progressing osteolysis, mostly of carpal and tarsal bones, leading to articular deformities and functional impairment, with or without nephropathy. The incidence has not yet been established. And there is no available treatment as of today

Objectives: To share the experience of MCTO identification in pediatric rheumatologist practice at federal center.

Methods: Totally 2 MCTO cases were identified in boys 6 ad 13 years old during the period 2009 – 2018. Standard rheumatological examination was performed. Genetic testing (Sanger sequencing) in 1 patient identified MAFB-gene mutation.

Results: Both patients had pain and swelling in wrist and ankle joints, flexion contractures in elbow joints, and gait abnormalities. Disease duration at the time of MCTO verification was 4 and 11 years. Both patients went through erroneous polyarticular JIA diagnoses. During the follow up patients’ ESR and CRP were normal, HLA B27 - negative, ANA and RF – negative. No visceral pathology, including kidneys, was found. Therapy included NSAIDs, glucocorticosteroids – in one case, MTX and Tocilizumab gave no effect in the second patient. Radiographic findings were severe osteolysis of wrist and feet bones. JIA diagnosis was ruled out and MCTO suspected, and later confirmed in both patients by a geneticist. Heterozygous MAFB gene c.206C>T (p.Ser69Leu) mutation was detected in one patient.

Conclusion: Specific phenotypical features and patient’s atypical status, osteolysis of the carpal and tarsal bones, absence of laboratory activity signs and of response to antirheumatic therapy is a sound motive to continue diagnostic elaboration in order not to miss rare genetically-linked conditions of the musculoskeletal system.

Disclosure of Interests: None declared


AB1046

PHYSICAL ACTIVITY ASSESSMENT IN CHILDREN WITH JUVENILE IDIOPATHIC ARTHRITIS COMPARED TO CONTROLS

Alia Fazaa, Meirem Sallam, Saudsen Miladi, Kmar Queninche, Leila Souabni, Selma Kassab, Selma Chekili, Leith Zakraoui, Kawther Ben Abdelghani, Ahmed Laatar. Mongi Sim Hospital, Rheumatology, Tunis, Tunisia

Background: Physical activity (PA), known to maintain optimal metabolic function and normal development could be impaired during Juvenile Idiopathic Arthritis (JIA).

Objectives: The aim of our study was to assess PA in children and adolescents with JIA compared to healthy peers using the physical activity questionnaire for children (cPAQ) and adolescents (aPAQ).

Methods: This is a cross-sectional study of measured level of PA in children and adolescents with JIA, compared to age and gender-matched healthy Tunisian schoolchildren. PA was estimated by cPAQ and aPAQ filled by the patient group and the reference group. If the child is unable or unsure to answer the questions we have helped with the parents response. The PAQ scores 2 as ‘low activity’, ≥2 and ≤3 as ‘moderate activity’, >3 as ‘high to vigorous activity’.

Results: A total of 55 patients (38 boys and 17 girls) with JIA and 60 healthy control schoolchildren were included. No significant difference in demographic background was found between the two groups. The mean age was 8.5 ± 4.12 years in the JIA group and 9.2 ± 3.51 years in the control group. Thirty-one patients (53%) had persistent oligoarticular JIA, 15 (27%) had polyarticular JIA, 5 patients (9%) had systemic JIA, and 4 (7%) had enthesitis-related arthritis. The median disease duration was 3.2 ± 2.8 years. The mean cPAQ was 2.101 ± 0.722 in the JIA group and 4.112 ± 0.644 in the control group (p=0.0001). Children and adolescents with JIA had a significantly lower levels of PA compared with their healthy peers as assessed by cPAQ/aPAQ (p=0.012). The time spent in each cPAQ/aPAQ item was significantly lower in the JIA group compared to the healthy group: sports activities (3.4 ± 0.5 versus 4.7 ± 1.4 hours/week, p=0.002), leisure time activities (2.2 ± 0.3 versus 6.2 ±1.3 hours/week, p=0.001), activities at school (1.1 ± 0.3 versus 2.1 ± 0.5 hours/week, p=0.001), and after school activities (0.5 ± 0.5 versus 2.5 ± 0.8 hours/week, p=0.001). Seventy six percent of the JIA group spent the day in the two lowest PA categories: sleeping and sitting, which was significantly higher compared with the reference group (p=0.001 and p=0.055, respectively).

Conclusion: In our study, children and adolescents with JIA were less physically active than the healthy peers as assessed by the PAQ. More objective methods are needed to better evaluate and quantify the PA.

Disclosure of Interests: None declared


REFERENCES


Disclosure of Interests: None declared


AB1045

MULTICENTRIC CARPOTARSAL OSTEOSLYsis (MCTO) IN PRACTICE OF PEDIATRIC RHEUMATOLOGIST: DIFFERENTIAL DIAGNOSIS WITH JUVENILE IDIOPATHIC ARTHRITIS

Svetlana Salugina1, Irina Nikiishina1, Olga Borodacheva1, Svetlana Rodionovskaya2, V.A.Nasonova Research Institute of Rheumatology, Moscow, Russian Federation; 1Central Children’s Hospital of Federal Medical Biological Agency of Russia, Moscow, Russian Federation

Background: Rare genetic pathologies involving the musculoskeletal system can be erroneously misdiagnosed as juvenile idiopathic arthritis (JIA).