Objectives: Describe two brothers affected by MCTO highlighting the importance of a genetic diagnosis.

Methods: B1 (20 years) presented age 3 years with swollen painful feet and restricted movement in his fingers and wrists and was diagnosed with polyarticular juvenile idiopathic arthritis (JIA). After no response to steroid treatment he developed rapid onset fixed flexion deformity of both elbows. He had mild learning difficulties. Based on clinical and radiological findings he was diagnosed with carpotarsal osteolysis age 4 years. Bisphosphonates and vitamin D replacement did not halt progressive bony destruction. Consistent with the literature osteolysis stabilised in the teenage years. Renal function remains normal.

B2 (5 years) is a dizygotic twin and younger sibling of B1. All other family members: parents (non-consanguineous), twin sister, sister, brother and half-brother are healthy. B2 presented aged 21 months with bilateral painful swollen wrists and reduced range of movement. His development was normal and comparable to his twin. Clinical presentation was similar to B1 who was yet to receive a genetic diagnosis. Plain films were not diagnostic as carpi unossified. Regular monitoring of renal function was advised although it was noted the nephropathy associated with MCTO does not normally manifest until the second decade.

At genetic review an autosomal recessive form of carpotarsal osteolysis was considered most likely. The elbow contractures in B1 were consistent with this phenotype although subcutaneous nodules were absent. Genetic testing for *MAFB* gene alterations identified an identical mutation in B1 and B2 (*MAFB* – c176c>Tp(Pro59Leu) confirming the autosomal dominant MCTO. B2 developed hypertension, heavy proteinuria and hypoalbuminaemia at 3 years, responding to tacrolimus.

Results: Two brothers with clinical features of MCTO and identical mutations in *MAFB* gene are described. The phenotype varied significantly with early renal disease in one and mild learning difficulties in the other. Inheritance of this autosomal dominant condition in these two brothers with unaffected parents could result from either gonadal mosaicism or incomplete penetrance. There is a case of incomplete penetrance in MCTO described in the literature. [2]

Conclusion: Individually skeletal dysplasias are rare yet as a group they represent an important differential to JIA requiring different management. Pursuing a genetic diagnosis in carpotarsal osteolysis is important not only for prediction of recurrence risk. It allows a definitive diagnosis to be made earlier than is possible clinically/radiologically. Significantly different phenotypes yet the same genetic mutation lends weight to the possibility of modifier genes or other epigenetic mechanisms/environmental factors affecting disease penetrance as proposed by Dworschak et al [2]. A greater understanding of these may enable prediction of renal involvement and aid development and stratification of new therapies.

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AB0952

RETROSPECTIVE STUDY OF PROCEDURAL PAIN ANALYSIS DURING JOINT INFILTRATION IN PATIENTS WITH JUVENILE IDIOPATHIC ARTHRITIS

Emanuele Cocchiara¹, Claudia Ferrari², Michela Cappella³, Carlo Salvarani¹, Alessandro De Fanti³. ¹Policlinico of Modena University Hospital of Modena, Rheumatology, Modena, Italy; ²Arcispedale Santa Maria Nuova, Rheumatology, Reggio Emilia, Italy; ³Arcispedale Santa Maria Nuova, Pediatrics, Reggio Emilia, Italy

Background: Juvenile Idiopathic Arthritis (JIA) is the rheumatic disease more common in childhood. Intra-articular corticosteroid injections (IAC) are a mainstay in the treatment of this disease, unfortunately it is often associated with pain and anxiety, for this reason is extremely important to pay attention both to procedural analgesia and to the reduction of anticipatory anxiety.

Objectives: The primary aim was evaluate the impact of the our analgesic procedure on the perception of infiltration pain; the secondary aims

were stratify the results according to the demographic characteristics, number of joints infiltrated for each patient.

Methods: 40 patients (pt) studied between august 2016 and august 2017 (1 year) received a IAC [F/M=25/15, age 8.63 ± 3.41 yrs, weight 32.1 ± 13.9 kg; monoarticular JIA 7 pt (17.5%), oligoarticular 19 (47.5%), polyarticular 14 (35%). 27/40 (67.5%) pt infiltrated in one joint, 13/40 (22.5%) in two or more joints].

One hour earlier of the procedure for each pt was applied 1% prilocaïne cream and 30 minuts before administered Midazolam orally at a dose of 0.5 mg/kg (maximum 15 mg), 50% nitrogen protoxide mixture dispensed with a mask, in spontaneous breathing and continuous flow. In 25/40 (62.5%) pt distraction techniques were performed during IAC and in 24/40 (60%) was also applied ice spray before IAC. To monitor pain, before and after IAC, appropriate scales have been used, stratified by age: CHIPPS scale (pt ≤ 5 yrs), Wong-Baker scale for ages 5 to 8, numerical scale analog visual VAS (pt ≥ 8 yrs). CHIPPS and VAS scales <4 : indicative of mild pain. For each pt pain assessment was performed before and after IAC.

Results: VAS before IAC = 0.90 ± 1.67 /Median (M) = 0, after IAC = 1.07 ± 1.56 /M = 0; CHIPPS before IAC = 0.82 ± 1.88 /M = 0, during IAC = 2.75 ± 3.01 /M = 2. No significant correlation between VAS scale scores and after IAC and of CHIPPS both before and during IAC and respectively of age, weight, sex, application of ice spray, application of distraction techniques before the execution of the procedure, number of infiltrated joints. Significant correlation between VAS before and after IAC (RHO Spearman 0.397, $p = 0.018$), CHIPPS before and during IAC (RHO Spearman 0.599, $p \leq 0.0001$). Highly significant correlation coefficient between VAS and CHIPPS before IAC (RHO Spearman 0.6, $p = 0.0001$) and between CHIPPS during IAC and VAS after IAC (RHO Spearman 0.403, $p < 0.05$). Significant difference between CHIPPS before and after IAC (Wilcoxon Ranks Signed Test, $R = 8.5$, $p < 0.0001$), not between VAS before and after IAC ($R = 83$, $p = 0.62$).

Conclusion: In our study the intensity of pain would seem exclusively subjective, in fact it does not depend: on age, number of IAC, on the application of distraction techniques and application of ice spray. Anyway the protocol that we used seems to be effective on pain control (VAS and CHIPPS scale respectively after and during IAC: $m \pm sd$ and M. <4 points) and it can be performed on outpatients safely without an anesthetic procedure. Our results encourage to expand the series with a future prospective trial.

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AB0953

EAR INVOLVEMENT IN PATIENTS AFFECTED BY JUVENILE IDIOPATHIC ARTHRITIS

Giovanni Conti¹, Giulia La Barbera¹, Carmela Visalli², Giuseppe Alberti³, Paola De Pasquale³, Francesco Borgia⁴, Francesco Ciodaro³. ¹AOU Policlinic G Martino, University of Messina, Pediatric Rheumatologic Unit, Messina, Italy; ²AOU Policlinic G Martino, University of Messina, Radiology Unit, Messina, Italy; ³AOU Policlinic G Martino, University of Messina, Otolaryngology Unit, Messina, Italy; ⁴AOU Policlinic G Martino, University of Messina, Dermatology Unit, Messina, Italy

Background: Few clinical studies in pediatric patients have shown a possible link between hearing loss and Juvenile idiopathic arthritis (JIA). It could be related to the involvement of the joints of the ossicular chain as a result of the autoimmune inflammatory process in the middle and inner ear

Objectives: The aim of this study was to assess the frequency of hearing impairment in patients affected by JIA versus a healthy control group.

Methods: We studied 64 ears of 32 JIA patients (25 girls and 7 boys, mean age 15 years, age range 8-19 years) from June to December 2018. The mean disease duration was 7 years (± 4 DS). Polyarticular JIA was diagnosed in 6 patients, oligoarthritis JIA in 19; 5 patients had psoriatic arthritis and 2 spondyloarthritis. The control group consisted of 60 ears of 30 healthy children, sex-and age-matched. Patients with previous otitis media at least 3 months and/or ear surgery were excluded. The study was approved by the local ethics committee and parents and/or patients signed their informed consent. All subjects underwent