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

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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

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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

audiological examination involving otomicroscopy, audiometry, tympanometry and stapedius reflex. Rheumatologic evaluation included joint examination with application of a measure of functional ability (disability) using the Childhood Health Assessment Questionnaire (CHAQ). The investigators were blinded about JIA type, severity, duration, and treatments of the patients.

Results: 11/32 (34%) JIA patients had abnormal audiological exam. Hypoacusia was significantly more frequent ($p<0.05$) in JIA patients (15/64 ears=23%) respect to the control group. In particular, hypoacusia was associated to psoriatic arthritis (6/15 ears). Conductive hearing loss was detected in 12/15 involved ears, while sensorineural type in 3/15. In the JIA group, according to the Jerger classification for tympanometry, abnormal findings were observed in 7 ears. The stapedius reflexes were absent in only one patient (3%). JIA patients with hypoacusia had a greater CHAQ (0.4+/-0.3) than JIA patients with normal audiometry (0.18 +/-0.29).

Conclusion: The presence of hearing impairment and/or abnormal tympanogram suggests early involvement of the tympanic-ossicular complex, especially in patients with psoriatic arthritis. These findings could be a marker of the JIA activity. Therefore, it is advisable that JIA patients perform periodic audiological examination to early detect eventual hearing impairment, in order to prevent hearing loss in adult age.

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Disclosure of Interests: None declared

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AB0954 PEDIATRIC RHUPUS A RARE OVERLAP SYNDROME: CASE SERIES OF MEXICAN CHILDREN'S HOSPITAL FEDERICO GOMEZ

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Background: Rhupus is an overlap syndrome of rheumatoid arthritis and systemic lupus erythematosus (SLE) in adult patients. Its pediatric presentation is very rare, the presentation of both juvenile idiopathic arthritis (JIA) and SLE as a clinical manifestation of the same patient has aroused different theories being the most accepted a true overlap between SLE and JIA. To diagnosis a patient with Rhupus present with clinical symptoms of SLE with positive ANA, antiDNA or antiSm, associated with clinical symptoms of JIA

Objectives: Describe clinical and serological findings of pediatric patients diagnosed with Rhupus of Mexican Children's Hospital Federico Gomez

Methods: We present a case series of 4 patients diagnosed with Rhupus. We describe the demographic characteristics, clinical presentation, serological finding, the criteria for the diagnosis of SLE (ACR 1997) and JIA (ILAR 2001) and the treatment installed

Results: We describe four patients with Rhupus, the medium age at diagnosis was 12.5 years, and it was more common in female 3:1. Three presented joints manifestations, integrating an initial JIA diagnosis. The evolution to an overlap syndrome took an average time of 19 months. The main symptom at diagnosis was erosive arthritis, poliarticular, morning stiffness and decreased joint range of motion. Three patients met 6 classification ACR 1997 criteria for SLE. All of the patients met ILAR 2001 criteria for JIA diagnosis. The four patients were treated with multitarget therapy. Joint manifestations were treated with methotrexate, and kidney and neurological manifestations with MFM and IV CFM. One patient required etanercept therapy for persistent joint disease

Abstract AB0954 Table 1. Demographic and clinical characteristics

	CASE 1	CASE 2	CASE 3	CASE 4
AGE AT DIAGNOSIS	15	11	11	13
SEX	F	M	F	F
FIRST DIAGNOSIS	SLE	JIA	JIA	JIA
TIME TO OVERLAP months	Simultaneous	18	15	36
SLE ACR 1997	7	6	6	6
JIA ILAR 2001	Si	Si	Si	Si

TYPE AIJ TREATMENT	Oligoarticular MTX 3 months CFM MFM (kidney disease) HCQ steroids	Polyarticular MTX 3 months MTX sulfasalazine 6 months MFM 6 months Etanercept HCQ steroids	Polyarticular MTX 3 months MTX sulfasalazine present HCQ steroids	Polyarticular MTX 6 months CFM MFM (neurological condition) HCQ steroids
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All of the patients presented with elevated acute phase reactants, positive ANA, antiDNA and antiCCP. Two patients had positive antiSm antibody and three presented with positive RF

Abstract AB0954 Table 2. Serological features

	CASE 1	CASE 2	CASE 3	CASE 4
ANA	PH1:1280	PH1:5120	PH1:640	PH1:1280
ANTI DNA	+	+	+	+
ANTI SM	+	-	+	-
VSG/PCR	+	+	+	+
FR	-	+	+	+
ANTI CCP	+	+	+	+
ANTI RO	-	-	+	-
AFLS	+	-	+	+

Conclusion: The coexistence of two or more connective tissue disease in the same patient is a very uncommon finding. The overlap of SLE and RA has been estimated between 0.01-2%. There are no reliable data about the overlap of SLE and JIA. Patients with JIA and positive anti-nuclear antibodies should have a thorough follow up taking into account the possibility of developing a juvenile Rhupus, evaluating cutaneous, renal, neuropsychiatric or hematologic manifestations in each visit. Its manifestations can be very devastating, increasing the morbidity and mortality of children. Previous findings showed that positive antiCCP antibodies are very specific for Rhupus diagnosis (96-98%). The clinical response to conventional therapy as not as than seen in patients with JIA, requiring of a multitarget treatment and biological agents to achieve a respond. Severe organ specific manifestations such as lupic glomerulonephritis and neuropsychiatric disease, are common. It is very important to recognize the Rhupus in order to make a prompt diagnosis and intervention that assures a better outcome for patients

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AB0955 TRANSITIONAL CARE: A SINGLE CENTER ITALIAN EXPERIENCE

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Background: Transitional care from paediatrics to adults is an important step in the growth of the adolescent patients. One of the final goals is to guarantee the continuity of health care, increasing compliance in treatment and visits.

Objectives: To describe disease features and adherence to the follow-up in rheumatic patients that were transferred from Paediatric Clinic to our adult department.

Methods: From May 2016 to May 2018 the transition has been performed with a visit in presence of both specialists and 28 patients were evaluated. For juvenile Idiopathic Arthritis (JIA) we registered: category of