Nephropathy onset (age; years)	3	9	None
Current state	Proteinuria (720mg/24h)	Proteinuria (1.3g/24h)	Normal kidney function
Serum C1q concentration	174 mg/l	312 mg/l	176 mg/l

Conclusion: C1q plasma levels in our patients were normal, suggesting that MCTO-associated genetic variants do not play a role in MafB-dependent regulation of complement component C1q production in humans. Further studies are necessary to exclude a role of complement system in the progressive nephropathy of patients with MCTO.

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AB1036 AN UNSOLVED CASE: IS THIS A CANDLE-LIKE SYNDROME?

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Background: Chronic Atypical Neutrophilic Dermatosis with Lipodystrophy and Elevated Temperature (CANDLE) syndrome is a complex autoinflammatory disorders arising from inborn defects in immunoproteasome. Several genes can be involved and cases with digenic inheritance have been described. However, many cases remain without the identification of a specific genetic defect. A positive interferon signature is typically found in patients and may serve as a diagnostic clue.
Objectives: To describe clinical and genetic features in a girl with CANDLE and in her relatives with a variety of different rheumatologic complaints.
Methods: We performed Whole Exome Sequencing (WES) on 10 family members. Moreover, we assessed RNA-seq on three sample from the proband, collected during acute phases of disease (samples positive to class I interferon signature (IS)) (1), and her parents. Results of RNA sequencing (RNA-seq) were compared with specimens from healthy controls.
Differentially expressed genes (DEGs) were filtered by fold change > 2 and padj < 0.05. DEGs enrichment were performed using different R packages, such as pathfindR.
Results: We describe the case of a 20 years old girl with clinical and biological data supportive of CANDLE syndrome. At the age of 3 years, she started presenting a clinical picture reminiscent of amyopathic dermatomyositis, with skin rash, lipodystrophy, subcutaneous panniculitis nodules, and more recently with chilblains, skin ulcerations, polyarticular arthritis and alopecia.
Her pedigree includes several relatives with rheumatic disorders, but none has a clinical picture as complex and severe as our patient. This girl was found to have a strongly positive class I IS in peripheral blood cells. After several unsuccessful therapeutic attempts with antirheumatic drugs and biologics, the girl showed a dramatic clinical response to the JAK inhibitors tofacitinbib.
IS resulted positive also in 4 of her relatives, three of whom presented also rheumatologic symptoms. Conversely, one uncle of the girl was affected with rheumatologic symptoms but had negative IS. The pedigree may suggest a complex pattern of inheritance, likely with a major dominant disorder, whose expression can be modulated by multigenic and/or environmental factors.
WES failed to detect significant genetic variants in proteasome components. However, RNA-seq revealed a profile of differentially expressed IFN-regulated genes similar to that reported by Anja Brehm et al. in subjects with CANDLE/PRAAS (2).
Conclusion: Our results suggest that our family may present a multigenic form of CANDLE, with a complete clinical picture only in the proband, whilst other relatives may only present partial or incomplete forms of the disease.

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AB1037 SEROLOGICAL PHENOTYPES, UVEITIS AND DISEASE ACTIVITY INTO ADULTHOOD: LONG TERM OUTCOME IN A COHORT OF PATIENTS WITH JUVENILE IDIOPATHIC ARTHRITIS

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Background: Deepening the long-term study in adulthood of Juvenile Idiopathic Arthritis (JIA) is fundamental in order to expand knowledge of the pathogenesis, optimize the therapeutic choices, favour a more active communication between paediatric and adult care specialists.
Objectives: The present project deals with adult patients affected by JIA. The main objectives were those of: analyse the serological profile (rheumatoid factor –RF- IgM, ACPA IgG, ANA) of such patients to investigate possible seroconversions in adulthood compared to the diagnosis in paediatric age; investigate whether correlations between antibodies (Ab) and diagnostic subgroups subsist in adulthood; evaluate the association between Ab and disease activity; investigate the association between the presence of uveitis in the medical history and specific Ab, diagnostic subgroups and disease activity.
Methods: 68 patients were selected. Data were collected from medical records, a sample was taken to search for ANA, RF and ACPA; clinical data were collected on the evaluation of disease activity, using JADAS27 and SDAI as clinical scores (fig.1)
Results: The data obtained were significant for the negativization of ANA in adulthood: at the diagnosis 45.6% of patients had ANA positivity, while in adulthood 13.2%. The difference was statistically significant. The Ab picture of both RF and ACPA remains unchanged in adulthood, therefore there was neither a significant positivization nor negativization. The concomitant positivity for RF and ACPA was found to exist, demonstrating statistical significance for both the diagnosis and the adulthood (p 0.05). A higher incidence of uveitis was not correlated either with the presence of ANA in paediatric age, nor in adulthood, but is instead associated with the diagnosis of oligoarticular JIA (p = 0.002). Analyses of the positivity for RF and ACPA (in relation to disease activity calculated with clinimetric indices such as JADAS27, SDAI and CDAI) detected the negative prognostic role of the two Ab as they correlated with higher disease activity in the population of the patients in the study (fig.2)
Conclusion: In a rheumatological scenario enriched of great improvements in the diagnostic and therapeutic field during the last decades, a far from negligible portion of patients with JIA requires the continuation of rheumatological care in adulthood. The analyses of the present study report a significant negativization of ANA, thus suggesting the need to confirm the presence of these Ab in adulthood, and the difficulty in recognizing the classifying role of such Ab. The serological profile of RF and ACPA remains unchanged in adulthood, their correlation with the polyarticular subgroup is maintained even in the long term and both correlate with greater disease activity in adulthood.

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Fig.1	*Demographic, clinical and serological characteristics of patients (n=68)*					
Female n (%)	54 (79,4%)					
Age (±d.s.)	28 anni (±10)					
Disease duration (±d.s.)	17 anni (±10)					
Uveitis	12 (17,6%)					
Diagnostic subgroups	**Sistemic JIA** n=10 (14,7%)	**Oligoarticular JIA** n=28 (41,2%)	**Poliarticular JIA** n=20(29,4%)		**Psoriasic JIA** n=3 (4,4%)	**Entesitic JIA** n=7 (10,3%)
			RF Positiv n=9	**RF Negativ** n=11		
Female n (%)	6 (60%)	13 (46,4%)	9 (100%)	9 (81%)	2 (67%)	2 (28,5%)
Uveitis	0	8 (28,6%)	0	2 (18%)	0	2 (28,5%)

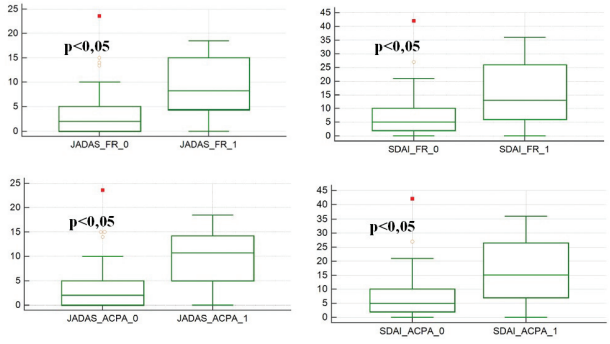

Figure 1

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AB1038 JUVENILE IDIOPATHIC ARTHRITIS INTO ADULTHOOD: HOW DO WE ASSESS DISEASE ACTIVITY?

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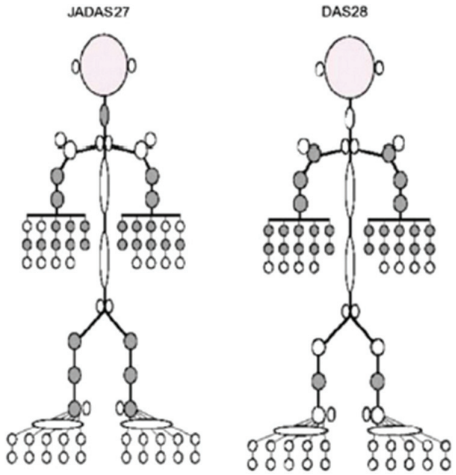
Background: Deepening the long-term study of Juvenile Idiopathic Arthritis (JIA) in adulthood is essential to increase the pathogenetic knowledge of the disease, to optimize the therapeutic choices accordingly, as well as to promote a more active communication between paediatric care and adult care specialists

Objectives: The present project, created as part of the "transition of care", aims to compare clinimetric scores of wide use for adult inflammatory rheumatisms of the adult (DAS28, CDAI and SDAI) with the JADAS27 score, which has been validated and widely used in order to quantify JIA's activities in the paediatric field. As of today, adult patients with JIA are usually evaluated with clinimetric scores developed for adult chronic rheumatic diseases (DAS28, CDAI, SDAI) and there is no consensus concerning which of these scores doctors should favour, so that the choice is quite autonomous and varies from centre to centre. It is therefore of interest to verify whether among these indices of purely rheumatological use of adults there is one that is more appropriate than JADAS27 which can be useful in monitoring adult patients with JIA.

Methods: The relevant clinical data were collected from 68 adult patients with JIA. A correlation analysis was performed between the clinimetric scores according to McNemar Test and Kappa by Cohen.

Results: The results obtained suggest that none of the clinimetric scale commonly used in the rheumatological clinical practice of adult patients can completely replace JADAS27. DAS28 is the score that goes further from an acceptable correlation with JADAS. Since both CDAI and SDAI are calculated with formulas that are similar to the one used for JADAS (algebraic sums of affected joints, subjective outcomes reported by the patient, clinical judgment of the physician), they happen to be a method of quantification of disease activity quite closer to JADAS itself. The analyses outlined a scenario in which a much larger portion of patients are classified in remission stages or in low disease activity when using CDAI and SDAI compared to JADAS27.



	Test di McNemar	Kappa di Cohen
JADAS-DAS28	p=0,00	0,08
JADAS-CDAI	p=0,00	0,11
JADAS-SDAI	p=0,00	**0,20**

Figure 1

Conclusion: This element inspired us to consider how in paediatric age a more "demanding" attitude towards the disease led to the validation of both a score and its very stringent cut-offs which are functional to a treat to target characterized by a complete remission whose main goal is to avoid long-term sequelae. SDAI was found to be the scale of common use in the adult care that more properly approaches the clinometry validated for the paediatric population (JADAS27). Although clinical common sense should not distract from assessing disease activity in this specific patient population from a global perspective, such a study could suggest using SDAI as clinimetric score of choice in adult patients with JIA. Further checks in larger population samples are obviously necessary.

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