Expressed constitutional disorders (fever, weight loss) observed in 13 pts (91.7%). 11 pts (78.6%) had acute cutaneous lupus, 5 pts (35.7%) – chronic cutaneous lupus, 3 (21.4%) – oral and nasal ulcers, 6 (42.8%) – nonscarring alopecia, 9 pts (64.3%) – polyarthritis, 2 pts (14.3%) – renal involvement, 2 pts (14.3%) – serositis, 1 (7.1%) – interstitial lung disease, 5 pts (35.7%) – neuropsychiatric disorder, including psychosis in 2 (14.3%). 4 patients had skin lesions atypical for SLE (1 - annular erythema, 1 - erythema nodosum, 2 - Ro-associated skin vasculitis). 12 pts (85.7%) had generalized lymphadenopathy. 12 pts (85.7%) had various hematological disorders: anemia – in 5 pts (35.7%), leukopenia – in 9 (64.3%), isolated lymphopenia in 1 (7.1%), thrombocytopenia – in 4 pts (28.6%). 13 pts had isolated involvement of salivary glands, 1 – combined with lacrimal glands. The decrease in salivary gland function was recorded in 50% of cases, hypolacrimia – in one case. Recurrent parotitis was present in only one case (7.1%). ANA were detected in 100% pts, anti-dsDNA – in 10 pts (71.4%), anti-Sm – in 7 pts (50.0%), anti-Ro – in 10 (71.4%), anti-La – in 7 (50%), RNP-70 – in 5 pts (35.7%), RF+ – in 6 pts (42.9%), hypocomplementemia – in 3 pts (21.4%). The most common was the combination of positive ANA, anti-dsDNA, antiRo with acute cutaneous lupus, polyarthritis, generalized lymphadenopathy and expressed constitutional disorders – 8 pts (57.1%). 4 pts (28.6%) had polyclonal hypergammaglobulinemia. 3 pts (21.4%) had concomitant autoimmune non-rheumatic disease; 1 - autoimmune hepatitis, 1 - type 1 diabetes mellitus, 1 - autoimmune thyroiditis. Median disease activity by SLEDAI at the time of jSLE verification was 11.5 scores [9.25;15.7].

Conclusion: According to our results, the frequency of detection of secondary SS in jSLE was higher than the literature data. The clinical features include a high frequency of constitutional disorders, lymphadenopathy, skin manifestations, high frequency of antiRo with a significantly lower incidence of kidney involvement, serositis than jSLE without SS. In pts with a diagnosis of SLE, the possibility of developing secondary SS should be considered (specially in girls with antiRo positive), the early detection of which affects the choice of therapy and prognosis.

References: [1]Malagón C, Gomez M, Mosquera C et al. Juvenile polyautoimmunity in a rheumatology setting. *Autoimmunity Reviews*, Volume 18, Issue 4, 2019, p 369-381. <https://doi.org/10.1016/j.autrev.2018.11.006>.

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AB0996 FAMILY CASE OF BLAU SYNDROME IN 3 CHILDREN, INITIALLY DIAGNOSED AS JUVENILE IDIOPATHIC ARTHRITIS

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Background: Juvenile Idiopathic Arthritis (JIA) is heterogeneous group of diseases which may include genetically determined conditions. Extremely rare monogenic hereditary autoinflammatory disease, such as Blau syndrome (BS) is usually difficult to recognize and JIA is initially established. BS is caused by a mutation in the NOD2/CARD5 gene and phenotypically characterized by triad of granulomatous polyarthritis, uveitis and skin rash.

Objectives: To present the family case of genetically confirmed BS in 2 siblings, initially diagnosed as patients with JIA.

Methods: Two brothers of 15 and 3 years old were examined in our clinic. Additional genetic assay was performed because of unusual clinical picture.

Results: *Case report.* Older brother, 15 y.o. developed arthritis of both wrist joints at the age of two. There was erythematous maculo-micropapular scaly rash on the trunk and extremities before the onset of arthritis. By 2009 (5 y.o.), the knees, ankles, and three PIP joints of the left hand were involved. Polyarthritis was characterized by severe effusion and periarticular tissues swelling. He was treated in regional hospital by NSAID, methotrexate, cyclosporine A without significant positive effect. Since 2012 etanercept was added for treatment with variable result. Inactive status of the disease is never reached. Repeated intra-articular GC injections were needed from 4 to 10 times per year. The patient was admitted to our clinic in November 2019. He was suffering from severe polyarthritis and uveitis de novo (granulomatous chorioretinitis) was detected. At the same time his younger 3 y. o. brother with recently started disease was hospitalized in our clinic. He had polyarthritis with typical features of "boggly" synovitis and tenosynovitis of wrists, ankles and knees, anterior uveitis was determined. The onset of arthritis was preceded by a small-spotted rash with desquamation. This classical clinical features in younger brother let us suggest the BS. There was no increasing of ESR and CRP in both sibs throughout the course of the disease. A molecular genetic study of the NOD2/CARD15 gene in both brothers revealed the same mutation of c.1001G>A (p. Arg334Gln). Because of the absence of specific treatment for this disease and due to insufficient effect of etanercept with uveitis de novo therapy was changed to golimumab and good initial effect is reached.

The youngest brother has started his therapy by methotrexate. It should be noted that the family has the eldest brother (20 y.o.), who has been suffering from arthritis since an early childhood with similar clinical picture. We are going to perform genetic analysis of the NOD2/CARD15 gene for the eldest brother.

Conclusion: Our clinical case shows that extremely rare BS may be misdiagnosed as JIA. Lack of efficacy of the etanercept therapy and uveitis de novo developing may be caused by genetic (*non-idiopathic*) nature of disease. Classic triad of boggy-arthritis, granulomatous uveitis and/or skin lesions without acute phase markers is required to perform genetic assay for the detection of a pathogenic mutation of the NOD2/CARD15 gene. This case is remarkable by the presence of BS in two (or 3) children of the family.

References:

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AB0997 IS HEIGHT ADJUSTMENT NECESSARY IN PEDIATRIC DENSITOMETRY IN ALL CHILDREN?

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Background: The current guidelines of the International Society for Clinical Densitometry (1) recommend that in children with linear growth or maturational delay, Z score results should be adjusted. Height for age Z score (HAZ) adjustment is valid and can be calculated using the formula the formula proposed by Zemel et al(2). It is possible that pediatric populations without linear growth or maturational delay, also benefit from HAZ, to prevent bone size from influencing the final Z score.

Objectives: To evaluate Z score variability adjusted and without adjusting for height for age.

Methods: We analysed data from densitometry performed on patients 2-20 years of age, from 2016 to 2018, assessed in the pediatric rheumatology office of our hospital for presenting risk factors for low bone mass/osteoporosis. The HAZ was calculated according to Zemel's formula.

Results: Data from 103 patients are presented. Its characteristics are summarized in Table 1

Table 1.

Mean age	9,8 years
Female	52,4%
Height Percentil ≤ 3	6,8%
Height Percentil ≥ 97	4,9%
LBM (Z score ≤ -2) spine	8,2%
LBM HAZ spine	6,4%
LBM whole body	10,5%
LBM HAZ whole body	7,2%

The table shows that the proportion of patients with BMD decreases in both the spine region and the whole body when adjusting for HAZ.

When evaluating the relationship between densitometric measurements we found that spine Z score (ZsS) and whole body Z score (ZsWB) had a correlation coefficient of 0,73 (p<0,001). There were no differences between their averages (p=0,170).

At the LBM cut-off point (Z score ≤ -2) there were discrepancies in 7%, where 5% presented LBM in ZsWB but not in ZsS. The concordance index at this point was 0,557.

When comparing these measures with their HAZ adjusted equivalents, we observe:

HAZ adjusted ZsS vs ZsS without adjusting: There were no differences between their averages (p=0,913) with a correlation coefficient of 0,78 (p<0,001). Concordance index at cut-off point for LBM was 0,498, with a discrepancy of 7%, where 2% had LBM according to HAZ adjusted ZsS, but not to ZsS without adjusting.

HAZ adjusted ZsWB vs ZsWB without adjusting: There were no differences between their averages (p=0,367) with a correlation coefficient of 0,82 (p<0,001). Concordance index at cut-off point for LBM was 0,557, with a discrepancy of 7%, where 2% had LBM according to HAZ adjusted ZsWB, but not to ZsWB without adjusting.

Conclusion: There are discrepancies at the LBM cut-off point depending on the HAZ adjustment.

The pediatric population without linear growth or maturational delay, can also benefit from HAZ adjustment, especially those with high height percentiles in which their size can hide a diagnosis of LBM.