Results: Demographics were 60.2±8.3 years of age, 65% female, BMI of 28.6±4.5 of kg/m²; 60% with Kellgren and Lawrence (KL) grade 2 and 40% with a KL grade 3. Semi-quantitative OMERACT ultrasound scores (Table.1) revealed good to excellent IRR (Kw=0.73 -0.88) for osteophyte, and moderate to good IRR (Kw=0.42-0.66) for synovitis. Conversely, quantitative measures of ultrasound pathologies had excellent IRR (ICC=0.84-0.95) except for synovial hypertrophy (ICC=0.67-0.72).

A significant association was found between semi-quantitative ultrasound synovitis and MRI effusion-synovitis (rs= 0.48, p=0.03). All three quantitative ultrasound measurements were significantly associated with quantitative scores of MRI (Table. 2).

In Bland-Altman plots for quantitative measurements, there were systemic offsets of 0.6 mm, 1.9 mm and 9.8 mm for osteophyte and meniscal extrusion and synovitis respectively (Fig. 1).

Conclusion: These is moderate to good IRR between operators with varying experience using the OMERACT knee scoring image atlas for osteophyte and synovitis. While quantitative ultrasound measurements showed excellent IRR and significant association with MRI quantitative outcomes, the absolute feature-specific agreement is called into question.

REFERENCES

- [1] Bruyn, G.A.W., et al., An OMERACT reliability exercise of inflammatory and structural abnormalities in patients with knee osteoarthritis using ultrasound assessment. *Annals of the Rheumatic Diseases*, 2016. 75(5): p. 842-846.

Disclosure of Interests: None declared

DOI: 10.1136/annrheumdis-2019-eular.2134

AB1168 SIGLEC1/CD169 IS A SENSITIVE MARKER FOR MONOGENIC INTERFERONOPATHIES

Banu Orak^{1,2}, Marc Nikolaus^{1,3}, Ellen Knierim^{1,3}, Angela Kaindl^{1,3}, Manuela Theophil⁴, Axel Panzer⁴, Barbara Zieba⁵, Frédéric Ebstein⁵, Elke Krüger⁵, Nadine Unterwalder⁶, Christian Meisel⁶, Tilmann Kallinich^{1,2}. ¹Berlin, Center for Chronically Sick Children (Sozialpädiatrisches Zentrum, SPZ) – Charité Universitätsmedizin Berlin, Berlin, Germany; ²Berlin, Department of Pediatrics, Division of Pneumology and Immunology with intensive Medicine, Charité Universitätsmedizin Berlin, Berlin, Germany; ³Berlin, Department of Pediatric Neurology, Charité – Universitätsmedizin Berlin, Berlin, Germany; ⁴Berlin, Pediatric Neurology, DRK Klinikum Berlin-Westend, Berlin, Germany; ⁵Greifswald, Institute of Medical Biochemistry and Molecular Biology, Universitätsmedizin Greifswald, Greifswald, Germany; ⁶Berlin, Department of Immunology, Labor Berlin GmbH, Berlin, Germany

Background: Monogenic Interferonopathies are a rare group of inflammatory diseases that are difficult to diagnose in the onset phase given the lack of well-defined disease-markers.¹ A correlation with interferon-stimulated genes (ISG) has been reported for *SIGLEC1* (syn. CD169), in systemic lupus erythematosus (SLE).² Furthermore, expression of *SIGLEC1* on monocytes is the second highest ISG in SLE.³

Objectives: To study the relevance of *SIGLEC1* as a putative diagnostic marker for early detection of interferonopathies.

Methods: Clinical data, classical inflammatory markers, blood count values and genetic information were obtained from the medical files of eight patients with genetically confirmed monogenic interferonopathies. *SIGLEC1* expression was measured by flow cytometry with a highly standardized quantitative assay with a reference range in healthy controls less than 2500 *SIGLEC1* molecules/monocyte. Additionally, transcriptional level of *SIGLEC1*, *IFI44L*, *IFI27*, *ISG15* and *RSAD2* as type I Interferon stimulated genes were assessed by real-time PCR.

Results: All eight patients with interferonopathies carried mutations in the genes *TREX1* (n=3), *IFIH1* (n=2), *SAMDH1* (n=2) and *RNASE2HB* (n=1). Mean age of patients was 12 years (range 6 months to 49 years). Six of eight patients showed neurological symptoms consistent with Aicardi-Goutières-Syndrome presenting developmental delay and microcephaly. Five patients showed abnormalities on cranial MRI including periventricular calcifications and corpus callosum thinning. Two patients were diagnosed with Singleton-Merten-Syndrome presenting abnormal ossification of extremities and dental anomalies. One patient with homozygous *TREX1* mutation presented with postnatal glaucoma, microcephaly, sensorimotor polyneuropathia and recurrent fever with persistent chilblain lesions. All patients had elevated *SIGLEC1* levels (mean molecules/monocyte +/- SD: 10272 +/- 3746) without high levels of standard inflammatory markers. In six patients, elevated *SIGLEC1* expression showed dysregulation of the type 1 interferon pathway prior to genetic testing. The relative

expression (Δ Ct) of all ISG's was significantly elevated in comparison to healthy controls (*SIGLEC1* p= 0,0167, *ISG15* p=0,0015, *RSAD2* p=0,0067, *IFI44* p=0,0001, *IFI27* p=0,0056).

Conclusion: We report high expression of *SIGLEC1* in monogenic interferonopathies like Aicardi-Goutières-Syndrome.

Therefore, *SIGLEC1* qualifies as an easy accessible and cheap diagnostic marker to screen patients with suspected interferonopathy.

REFERENCES

- [1] Crow, Y.J., *Type I interferonopathies: mendelian type I interferon up-regulation*. *Curr Opin Immunol*, 2015. 32: p. 7-12.
- [2] Rose, T., et al., *Are interferon-related biomarkers advantageous for monitoring disease activity in systemic lupus erythematosus? A longitudinal benchmark study*. *Rheumatology (Oxford)*, 2017. 56(9): p. 1618-1626.
- [3] Biesen, R., et al., *Sialic acid-binding Ig-like lectin 1 expression in inflammatory and resident monocytes is a potential biomarker for monitoring disease activity and success of therapy in systemic lupus erythematosus*. *Arthritis Rheum*, 2008. 58(4): p. 1136-45.

Disclosure of Interests: Banu Orak: None declared, Marc Nikolaus: None declared, Ellen Knierim: None declared, Angela Kaindl: None declared, Manuela Theophil: None declared, Axel Panzer: None declared, Barbara Zieba: None declared, Frédéric Ebstein: None declared, Elke Krüger: None declared, Nadine Unterwalder: None declared, Christian Meisel: None declared, Tilmann Kallinich Grant/research support from: Novartis, Speakers bureau: Sobi, Roche, Novartis, CLB

DOI: 10.1136/annrheumdis-2019-eular.5506

AB1169 ASSOCIATION OF ANXIETY, DEPRESSION WITH ULTRASONOGRAPHIC EVALUATION OF THE JOINTS, DISEASE ACTIVITY, FUNCTIONAL DISABILITY AND QUALITY OF LIFE IN PATIENTS WITH RHEUMATIC DISEASES:

Rita Osipiants¹, Victoria Nadocheeva², Madina Bogdanova³, Marina Kanevskaya⁴, Temuri Mirilashvili⁵. ¹Department of Internal Diseases, RUDN University; Eramishantsev City Clinical Hospital, Moscow, Russian Federation; ²Department of Internal Diseases, RUDN University, Moscow, Russian Federation; ³FSBI "Polyclinic N21" of the Administrative Department of the President of the Russian Federation, Moscow, Russian Federation; ⁴Sechenov Moscow Medical University, Moscow, Russian Federation; ⁵Eramishantsev City Clinical Hospital, Moscow, Russian Federation

Objectives: The aim of this study is to evaluate anxiety and depression disorders and to study their correlation with disease activity, ultrasonographic (US) finding indicative of synovitis, functional status and quality of life in patients (pts) with rheumatic diseases.

Methods: 39 pts (F/M=29/10) with rheumatoid arthritis (RA, n=17), ankylosing spondylitis (AS, n=10), psoriatic arthritis (PsA, n=12) were included. All RA pts fulfilled the ACR/EULAR 2010, AS pts - the ASAS, PsA pts - the CASPAR criteria. 20 pts (8 RA, 6 AS, 6 PsA) were performed US examinations (Mindray DC-N6 (China), C5-2, L10-13 MHz probes) included bilateral of the hip and knee joints. Each joint was scored according to the OMERACT definitions of pathology. Functional status (BASFI), disease activity indices (DAS28-CRP, ASDAS-CRP, DAPSA), anxiety and depression levels (Hads-A, Hads-D) and quality of life (RAPID3, EQ-5D) were assessed. The Mann-Whitney U-test was applied for intergroup comparison and correlation was evaluated using a Spearman's Rank two-tailed test (R value is shown).

Results: Median age of 39 pts - 52 (44;61) years and the body mass index - 28 (23;30). Mean anxiety index was 6.7±3.4, depression index - 6.6±3.3. Anxiety disorders were detected in 13/39 pts (33.3%), depression disorders in 17/39 pts (43.5%). Pts with Hads-D ≥8 (n=17) had higher score ASDAS-CRP (4.6 (4.2;5.0) vs 3.1 (2.7;3.9), p<0.01), BASFI (7.2 (6;7.3) vs 4.2 (3;5.5), p<0.05), EQ-5D index (9.8 ±1.2 vs 8.5±1.6, p<0.01) and the same tendency to US score and DAS28-CRP compared pts without depression. But pts with Hads-A ≥8 (n=13) had only higher EQ-5D (10 ±1.2 vs 8.7±1.6, p<0.005) and RAPID3 (17.1 ±5.8 vs 15.6 ±1.6, p<0.05). However, US - synovitis of the hip joints was positively correlated with Hads-D (r=0.713, p<0.05) and Hads-A (r=0.587, p<0.05).

Conclusion: Anxiety and depression more common in patients with active rheumatic diseases. Psychological disorders are associated with disease activity, US- finding of synovitis and the severity of functional disability.

Disclosure of Interests: None declared

DOI: 10.1136/annrheumdis-2019-eular.8344