1. Streszczenie pracy w języku angielskim

Rheumatic diseases cover a broad spectrum of disorders characterised by the presence of inflammatory changes within the connective tissue. Establishing an accurate diagnosis of rheumatic disease is challenging due to the need for obtaining a detailed medical history and physical examination, as well as a panel of laboratory tests and imaging procedures. Magnetic resonance imaging (MRI) of the musculoskeletal system is one of the most important medical imaging techniques used in the diagnosis of rheumatic diseases, both in children and adults. Performing a whole-body or specific joint MRI in a patient allows a detailed assessment of the inflammatory process. The most commonly used sequences are T1-, T2- and PD-weighted with or without fat suppression (FS) and STIR/TIRM (short tau inversion recovery/turbo inversion recovery magnitude) sequences, as well as available post-contrast sequences. By performing musculoskeletal MRI examinations on patients, an appropriate diagnosis can be made and the course of the disease can be monitored. The increased number of MRI examinations being performed allows the development of more and more artificial intelligence algorithms that facilitate pathological lesion detection.

The aim of this dissertation is to evaluate the role of musculoskeletal MRI in the diagnosis of rheumatic diseases by:

- conducting a review of current guidelines and publications on the use of MRI in the diagnosis of juvenile rheumatic diseases;

- performing a detailed evaluation of lesions detected on whole-body MRI in patients with suspected juvenile idiopathic arthritis (JIA), juvenile idiopathic inflammatory myopathies (JIIM) and chronic non-bacterial osteomyelitis (CNO);

- to analyse the relevance of performing MRI of the sacroiliac joints (SIJ) in patients diagnosed with enthesitis-related juvenile idiopathic arthritis or juvenile psoriatic arthritis;

- to test for clinical utility a fully automated algorithm designed to detect inflammatory lesions on the SIJ MRI in patients with suspected axial spondyloarthropathy (axSpA);

- a case report of a patient with Ehlers-Danlos syndrome (EDS), chronic non-bacterial osteomyelitis and overlapping sacroiliac arthritis, in whom performance of sacroiliac joints MRI allowed an early diagnosis to be made and whole-body MRI was used to monitor treatment efficacy;

- a case report of a patient with seronegative juvenile idiopathic arthritis with polyarticular onset, in whom an MRI scan of the sacroiliac joints was performed to assess treatment efficacy.

Article 1

This review paper summarises the role of musculoskeletal MRI in the diagnosis of rheumatic diseases in paediatric patients based on an analysis of current guidelines from international rheumatology societies (ILAR - International League of Associations for Rheumatology, EULAR - European Alliance of Associations for Rheumatology, ASAS - Assessment in SpondyloArthritis, OMERACT - Outcome Measures in Rheumatology). The diagnostic criteria for juvenile idiopathic arthritis, chronic non-bacterial osteomyelitis, juvenile idiopathic inflammatory myopathies, juvenile systemic scleroderma and juvenile systemic lupus were reviewed comprehensively in terms of the clinical utility of MRI. The development of artificial intelligence methods to facilitate and reduce the description time of imaging examinations was also highlighted.

Article 2

The inclusion criterion for the study was performance of a whole-body MRI examination in a patient due to suspected rheumatic disease. Clinical presentation, laboratory findings and lesions described on whole-body MRI were analysed. Thirty-three patients with a diagnosis of JIA, JIIM, CNO and other diseases upon performing the above examinations were included in the study. The most common clinical manifestation reported by patients was chronic pain and morning stiffness. On physical examination, one in two patients had skin lesions and one in three showed decreased muscle strength. Laboratory tests revealed increased inflammatory parameters in four patients and elevated creatine kinase (CK) levels in six patients. Positive antinuclear antibody (ANA) titres were obtained in eleven patients, human leukocyte antigen B27 (HLA-B27) in five patients and human leukocyte antigen Cw6 (HLA-Cw6) in four patients. The most commonly described lesions on whole-body MRI were multifocal myositis, increased synovial fluid and bone marrow edema. There was a statistically significant correlation between myositis and the diagnosis of JIIM ($p < 0.05$).

Article 3

A total of 152 patients who underwent MRI of the sacroiliac joints for suspected JIA were included in this study. The descriptions of the above examinations, clinical observations and laboratory results were analysed, with particular focus on the presence of elevated inflammatory parameters, ANA, rheumatoid factor (RF), HLA-B27 and HLA-Cw6. The majority, that is 59.87%, of the patients met the criteria for the diagnosis of JIA according to ILAR. The most commonly described lesion on SIJ MRI was bone marrow edema, which was present in 36 patients (23.68%). SIJ MRI also showed the presence of joint stenosis in 10 (27.78%) patients, and erosions and synovial fluid equally in seven (19.44%) cases each.

Based on the diagnosis of JIA and bone marrow edema on SIJ MRI, the patient population was divided into three groups, the first comprising patients with JIA and bone marrow edema on SIJ MRI, the second comprising the remaining patients with JIA and the third comprising patients with negative observation for JIA. Patients overwhelmingly (84.21%) reported chronic pain as the main symptom, which in 31.25% involved the spine. Limitation of joint mobility was observed in 40.13% patients. Inflammatory parameters such as leukocytes, C-reactive protein (CRP), and erythrocyte sedimentation rate (ESR) were elevated in one in five patients. Positive ANA was found in 40 patients (26.32%), HLA-B27 in 60 (39.47%) and HLA-Cw6 in 29 patients (19.08%), while RF was found in only one (0.66%) patient. Statistically significantly more ANA-positive patients were in the second group.

Article 4

This study evaluates an already existing algorithm used for the detection of bone marrow edema in terms of its effectiveness depending on the technical correctness of the SIJ MRI. A total of 173 SIJ MRI examinations performed in patients with suspected axial spondyloarthropathy aged between 18 and 86 years were included in the analysis. The study consisted of six steps. In the first step, the correctness of the SIJ MRI examination was assessed by measuring the deviation angle, which was determined between the fully correct line drawn along the posterior edges of the first and second sacral vertebrae and the line delineating the actual plane in which the cross-sections were acquired in T1 and STIR sequences. The algorithm was then adapted to the latest guidelines defining when the presence of SIJ inflammation can be described on MRI. In the next step, the researchers performed manual segmentation of the sacrum and iliac bones to establish a reference to which the performance of the algorithm could be compared. Subsequently, the accuracy of the manual segmentations and the ones created by artificial intelligence was assessed. In the fourth step, the researchers assessed the presence of SIJ inflammation in the MRI using the SPARCC scale (Spondyloarthritis Research Consortium of Canada). Further, manual and automated segmentations of bone marrow edema in SIJ MRI were compared. In the final stage of the study, a statistical analysis of the bone and bone marrow edema segmentations was performed, as well as the results of the visual comparison of the two segmentations and the SPARCC scores. In particular, the focus was on verifying the differences between the groups to which the examinations were assigned according to the angle of deviation. The measurements revealed a full correctness of 20% of the SIJ MRI. The angle of deviation ranged from 0 to 29.2 degrees. The performance of the algorithm appeared to be similar regardless of the angle of deviation, demonstrating that it is clinically feasible to use artificial

intelligence to detect inflammatory lesions in SIJ MRI. The results of the bone marrow edema segmentation for all MRI included in the study were as follows: sensitivity 0.76, specificity 0.97, accuracy 0.97. In addition, the sensitivity for the recognition of single inflammatory lesion was calculated, with a value of 0.75 for all MRI examinations.

Article 5

The case report concerns a 17.5-year-old female patient treated for chronic non-bacterial osteomyelitis who was admitted to the Clinical Department of Pediatrics and Rheumatology of St. Louis Regional Specialised Children's Hospital in Cracow because of pain in the scapula, thoracic spine, shoulders and iliac joints. During the physical examination, knee and elbow joint hyperextension, excessive mobility of the phalanges, increased range of motion in the iliac joints with normal muscle strength and tension, and soft, doughy skin, with the presence of difficult-to-heal scars were observed. Laboratory results showed slight leucocytosis and normal CRP. To extend the diagnosis, the patient underwent MRI of the thoracic spine and sacroiliac joints. The examinations described foci of bone marrow edema in the Th3, Th4 and Th7 vertebral bodies and the head of the seventh rib on the left side and bilaterally in the sacrum, and the right iliac bone in the area adjacent to the SIJ. In addition, there were areas in the sacrum and iliac bones corresponding to foci of metaplasia of adipose tissue. Based on the above clinical signs and examinations, the patient was diagnosed with the classic form of Ehlers-Danlos syndrome and sacroiliac arthritis overlapping CNO.

Article 6

This publication describes the case of a patient aged 5 years and 2 months suffering from seronegative juvenile idiopathic arthritis of polyarticular onset with HLA-B27 presence. The first symptoms appeared at the age of 14 months and included soreness and swelling of the interphalangeal joints II-IV of the right hand and I-IV of the left hand. In September 2021, on admission to the Clinical Department of Pediatrics and Rheumatology of St. Louis Regional Specialised Children's Hospital in Cracow the patient reported soreness of the II and III proximal interphalangeal joints of the right hand and II-IV of the left hand. Physical examination revealed swelling of the above joints with preserved active and passive mobility and normal muscle tension. Laboratory tests did not indicate an increase in inflammatory parameters such as leukocytes, CRP or ESR. During hospitalisation, a computed tomography (CT) of the sacroiliac joints was performed, describing bilateral inflammatory changes. Therefore, it was decided to include methotrexate at a dose of 15 mg per week and sulphasalazine at a dose of 500 mg per day. During subsequent hospitalisations, treatment was

modified by adding the biologic drug - adalimumab achieving low disease activity. A follow-up MRI scan of the sacroiliac joints was performed in February 2024, which showed no areas of bone marrow edema, sclerosis or fatty remodelling in the subchondral layers of the joints. Laboratory tests did not reveal any elevation of inflammatory parameters at that time. Physical examination revealed only widened contours of the thumb and fingers III-IV of the left hand and fingers II-IV of the right hand, without the presence of pain, with preservation of active and passive mobility. Due to the established stationary condition, the existing treatment was maintained.

In conclusion, MRI of the musculoskeletal system is one of the key diagnostic methods in rheumatology. It allows a broad assessment of the inflammatory process at an early stage allowing appropriate treatment to be initiated and achieving rapid remission. Whole-body MRI is being performed increasingly often, especially in children, allowing to find foci of muscle edema, bone marrow edema and increased synovial fluid throughout the body in a single procedure. The examination is particularly important in patients with suspected juvenile idiopathic arthritis, juvenile idiopathic inflammatory myopathies and chronic non-bacterial osteomyelitis. Targeting the diagnosis of enthesitis-related juvenile idiopathic arthritis or juvenile psoriatic arthritis is possible by performing MRI of the sacroiliac joints in patients. This type of medical imaging should be considered especially in patients reporting chronic back pain. As the number of sacroiliac MRI examinations performed increases, so does the need to optimise the analysis time. This is made possible by the introduction of artificial intelligence methods into clinical practice, which will facilitate the work of radiology specialists by indicating potential lesions. The automated algorithm developed and validated in a clinical environment is a valuable tool with high specificity and accuracy in the detection of bone marrow edema in SIJ MRI in adults. However, the competence of the above algorithm should be expanded so that it can also be applied to the paediatric population. The practical usefulness of musculoskeletal MRI in the diagnosis and monitoring of the effectiveness of treatment of rheumatic diseases in the paediatric population is demonstrated on the example of the clinical cases presented.