Background: Osteochondrodysplasia are a group of hereditary disorders in which bones and cartilages grows abnormally. There is a great clinical variability between bone/cartilaginous dysplasias. Spondyloepiphysial dysplasia is one subtype that causes platyspondyly and widening of epiphyses, causing disproportionate short stature and early osteoarthritis.
We describe family members with a unique phenotype: normal stature, shortening of the fourth metatarsal, positive inflammatory tests and ultrasonographic findings similar to rheumatoid arthritis. The condition was inherited as a dominant trait. Marik, I et al. [1] described a family with similar manifestations - a pseudorheumatoid form, but without inflammatory findings.
Case series: Twelve patients from the same family with spondyloepiphyseal dysplasia were evaluated. All our patients had normal height. Ten patients presented with mechanical hand symptoms. Clinical synovitis of the hands (wrists and metacarpophalangeal) was identified in six patients. Five patients presented with synovitis by ultrasonography. Eight patients presented increased inflammatory tests (C-reactive protein and erythrocyte sedimentation rate). Rheumatoid factor and anti CCP were negative in all patients.
Conclusions: This family presents a similar phenotype to a spondyloepiphyseal dysplasia subtype called progressive pseudorheumatoid dysplasia associated with hypoplastic toes.
This phenotype was described in only 11 families, without publications in Brazil. However, a different feature in our patients is an inflammatory component, with high inflammatory tests and synovitis, turning into an important differential diagnosis of rheumatoid arthritis.
The mutation described in patients with pseudorheumatoid dysplasia is present in the COL2A1 gene, which encodes type 2 collagen.
The family reported in this study have not had the mutation identified yet. Our research aims to identify if we are facing a new mutation or if it is a phenotypic variation of the same mutation.

References

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SPOROTRICHOSIS PRESENTING AS A RHEUMATOID SYNDROME: A CASE REPORT

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Background: Sporotrichosis is a subacute to chronic infection caused by the dimorphic fungus Sporothrix schenckii. Clinical manifestations most commonly include cutaneous and lymphocutaneous forms of the infection. Occasionally the infection involves the joints, most commonly the knee. Rare forms of sporotrichosis include osteomyelitis occurring after local or hematogenous spread. The diagnosis of sporotrichosis is established by culture or histopathology findings. In cases of joint involvement, synovial fluid parameters often show relatively low white cell counts with a neutrophil predominance. We report a case of disseminated sporotrichosis presenting as a rheumatoid syndrome with characteristic skin lesions.
Case Report: A 56-year-old African-Brazilian woman, with history of kidney transplantation, presented to the emergency department because of a 4-month complaints of arthralgia, cutaneous lesions, weight loss and fatigue. Physical examination on presentation revealed polyarthritis of elbows, wrists, metacarpophalangeal and proximal interphalangeal joints, also multiple hyperkeratotic and hyperpigmented plaques. She had no history of fever or lymphadenopathy. Laboratory evaluation showed a white blood cell count of 6140 cells/mm3, erythrocyte sedimentation rate (ESR) of 120mm/h, C-reactive protein (CRP) of 118mg/L; rheumatoid factor (RF) and anti-citrullinated protein antibody (ACPA) were negative. Radiographs of hands and elbows were normal, though magnetic resonance of these joints demonstrated synovitis with small joint effusion and intense bone marrow edema, suggestive of septic arthritis and osteomyelitis. Cutaneous biopsy revealed granulomatous inflammation. Fungal culture of skin lesions showed Sporothrix schenckii. Bacterial and acid-fast bacilli (AFB) cultures were negative. US-guided arthrocentesis could not retrieve synovial fluid. The patient was treated with intravenous amphotericin B for 2 weeks and then with oral itraconazole. Because there was significant clinical and laboratory improvement, no surgical debridement was needed.
Conclusion: We reported an unusual case of osteoarticular sporotrichosis, presenting as a rheumatoid syndrome. Fungal infections, including sporotrichosis, should be considered as a differential diagnosis especially in immunosuppressed and alcoholics patients.

Consent for publication

The authors declare that they have obtained informed written consent from the patient's tutors for publication

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STUDY OF CARDIOVASCULAR REPERCUSSIONS IN CHIKUNGUNYA FEVER IN PATIENTS OF A UNIVERSITY HOSPITAL

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Background: Chikungunya fever (CF) is a seasonal infection caused by arbovirus Chikungunya (CHIKV). It can affect cardiac tissue and promote arrhythmias, hemodynamic instability, myocarditis, pericarditis, heart failure, coronary ischemic disease and dilated cardiomyopathy. The objective of this study is to evaluate the prevalence, clinical and epidemiological data of the cardiological alterations found in patients diagnosed with CF attended at a university hospital.
Methods: This is a descriptive, observational and cross-sectional study. It is based on the information contained in medical records and epidemiological notification sheets from January 2016 to January 2018. The studied population is the patients with laboratory confirmation for chikungunya fever, above 18 years of age, resident in Paraíba and that do not present coinfection with other arboviruses. During the collection, a survey of all reported cases of chikungunya in the hospital was carried out, then cases from other states, younger than 19 years and cases that did not have laboratory confirmation were excluded. Such information was used to build the database in Excel spreadsheets. Statistical analysis was performed using statistical software Statistical Package for Social Science (SPSS).
Results and conclusions: The research is still going on, and partial results have shown that of the 143 cases reported at Lauro Wanderley University Hospital, 45 cases were laboratory confirmed for Chikungunya. Of these, 6.67% of the individuals, all of them female, presented cardiovascular alterations with lipotimia and transient arrhythmias in the acute or subacute phases of the CF, requiring 01 hospital admission. Therefore, it is not possible to reach conclusions based only on these partial results, but with this sample, it is predicted that the prevalence of cardiovascular repercussions in chikungunya fever is low, and requires larger studies to conclude data on the prevalence of cardiovascular manifestations in patients of CF.

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STUDY ON THE EFFECTS OF STRENGHT TRAINING IN PAIN AND DISEASE ACTIVITY IN WOMEN WITH PRIMARY SJOGREN'S SYNDROME

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