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DEL(18)(Q22): A CASE REPORT

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BACKGROUND

Deletions of chromosome 18 are among the most common autosomal abnormalities compatible with life. Of these abnormalities, deletions from the long arm in the terminal portion are the most common. Affected individuals may experience a variety of signs and symptoms, including intellectual disability, short stature, hypotonia, genital abnormalities and feet deformities. Arthropathies related to 18q deletion syndrome have been rarely described.

CASE REPORT

Patient, 13 years old, karyotype 46, XX, del(18)(q22), was referred with a complaint of progressive articular deformities in hands and feet since 8 years of age. At physical examination, articular deformities were observed in both hands and feet, decreased range of motion (ROM) in elbows and left wrist, Raynaud's phenomenon and livedo reticularis more prominent in lower limbs. Morning stiffness, edema or pain were denied by patient and parents. Anti-CCP, ANA and rheumatoid factor (RF) were positive, and IgA levels were normal. Patient was diagnosed with juvenile idiopathic arthritis (JIA) and was prescribed methotrexate and hydroxychloroquine. Patient evolved with resolution of the limitation of ROM and halted progression of deformities.

CONCLUSION

At the present time, there are six case reports of seven patients with JIA related to 18q deletion syndrome. Of all reports, there are only two male patients. Age of onset varies from 4 to 13 years old. From all eight patients, four presented the oligoarticular form. Most commonly affected joints are knees, followed by ankles and small articulations of hands and feet, wrists and hips. Our patient is the only who did not present arthritis in the knees, was the first to present arthritis in the elbows, and also was the first one with RF positive. Unlike most of the reported patients, our patient did not present relevant pain complaints and presented Raynaud's phenomenon and lived reticularis. This case reinforces the importance of careful investigations in patients with 18q deletion syndrome, especially those with progressive joint deformity.