A case of neurofibromatosis presenting with Jaccoud arthropathy

Jaccoud Arthropathy (JA) was originally described with acute rheumatismal fever and subsequently associated with other rheumatological diseases. In JA, hyperextension develops in proximal interphalangial joints, and hyperflexion in distal interphalangial joints. JA may occur idiopathically as well as being associated with rheumatological diseases, lung malignancy, infections (HIV) and Parkinson's disease. Neurofibromatosis is a genetic childhood disease which presents with skin, eye, neurological and musculo skeletal system manifestations. So far, no case of coexistence of neurofibromatosis and Jaccoud arthropathy has been reported. This case is presented due to establishment of Jaccoud arthropathy in a patient with the diagnosis of neurofibromatosis.

Keywords: jaccoud arthropathy ● hand deformity ● neurofibromatosis

Introduction

Jaccoud Arthropathy (JA) was originally described with acute rheumatismal fever and subsequently associated with other rheumatological diseases. In JA, hyperextension develops in proximal interphalangial joints, and hyperflexion in distal interphalangial joints. JA may occur idiopathically as well as being associated with rheumatological diseases, lung malignancy, infections (HIV) and Parkinson's disease. Neurofibromatosis is a genetic disease of childhood presenting with dermatological, neurological, ophthalmological and musculo skeletal system findings.

Neurofibromatosis type-1 (NF1, von Recklinghausen disease) is a congenital neurocutaneous syndrome that can involve nervous system, bone, endocrine glands and other organs. NF type 1, occurs once every 2500 to 3000 births and defect at 17q11.2 gene has been incriminated for this disease. Its cardinal findings are; neurofibromas, 'cafe au lait' spots, axillary -inguinal freckles and iris hamartomas (Lisch nodules). 30% to 50% of neurofibromatosis type 1 cases develops due to spontaneous mutation. The rest are autosomal dominant. Patients are diagnosed in childhood especially with cafe au lait spots on skin and neurofibromas, which are the earliest signs. Diagnosis is made when at least two of diagnostic criteria developed in 1988 and updated in 1997 by NIH consensus conference are met. In NF1, skeletal system abnormalities such as osteopenia, scoliosis, sphenoïd dysplasia, congenital tibia dysplasia and pseudoarthrosis may occur. Cafe au lait spots are the earliest and most consistent symptoms. Neurofibromas commonly emerge on the body after the age of 10. In NF1, Lisch nodules occur at the rates of 10%, 50% and 100% respectively around the ages of 6, 30 and 60. One of the clinical characteristics of Neurofibromatosis type 1 is that the rate of benign and malignant tumors increase. To our knowledge, so far no case of coexistence of neurofibromatosis with Jaccoud arthropathy has been reported. This case is presented due to the establishment of JA in a patient with the diagnosis of neurofibromatosis.

Case

A 24-year-old female patient with known Neurofibromatosis type-1 (NF1) diagnosis, presented to rheumatology outpatient clinic with complaints of deformity in her fingers. It was learned that finger deformity started two years ago and progressed in time. It was not
accompanied by pain and stiffness. In family history, her
father had a diagnosis of NF1. In physical examination,
no pathology was found in vital findings and systemic
examination but ‘cafe au lait’ spots were observed in
axillary region and trunk and neurofibroma was detected
behind the ear (Figure 1). There was no arthritis in
musculo-skeletal system examination. Z deformity of
thumbs, swan neck deformities in distal interphalangeal
joints, decrease in flexion in metacarpophalangeal joints
were present, suggesting JA (Figure 2). Bilateral hand
graphy corroborated non erosive disease. In addition,
patient had scoliosis as well. In eye examination, Lish
nODULES were detected. Laboratory examination results
were as follows: Leucocyte:7200 K/Ul, neutrophil: 4200
K/Ul, hemoglobin: 13.1 gr/dl, thrombocyte: 247.000
K/Ul, creatinine: 0.4 mg/dl, CRP: 1mg/L (N:0-8),
sedimentation: 2 mm/h. Among autoimmune markers,
ANA, RF and anti –CCP were found to be negative.
Anti-HIV was negative. In bilateral hand graphy, no
erosion was detected and with thoraco-lumbar graphs,
scoliosis was confirmed. Orthopedic surgery was
planned due to scoliosis. Patient was kept under clinical
follow up.

Discussion

JA, is a clinical condition characterized by reversible
joint deformities such as swan neck, thumb subluxation,
ulnar deviation, 'boutonniere' and hallux valgus along
with lack of joint erosions in plain graphy. It was first
defined by François-Sigismond Jaccoud in 1869 in
a young patient with rheumatismal fever and chronic
joints deformities. JA has been described to occur
in association with some other disorders, especially
Systemic Lupus Erythematosus (SLE). Although less
commonly, it has also been associated with scleroderma,
Sjögren's syndrome, vasculitis, psoriatic arthritis, reactive
arthritis, ankylosing spondylitis, calcium pyrophosphate
deposition disease among rheumatological diseases and
was detected even in normal individuals. JA may also be
associated with lung malignancy, inflammatory bowel
disease, infections (HIV), and Parkinson's disease [1-5].
In a study carried out in Brazil in 2006, JA incidence
was found to be 2.8% in SLE [6]. In a different case
series, the incidence of Jaccoud arthropathy was found to
be % 3.47 in SLE patients [7]. JA is marked by a
deforming arthropathy which can mimic clinical presentataion of RA: Although JA may involve all joints,
it mostly presents with ulnar deviation, swan neck and
boutonniere deformities, and Z-deformity of thumb,
more than one non erosive subluxation along with
serious deformations in hands. Subluxation in all MKP
joint and ulnar deviation is usually the first sign and
swan neck and boutonniere and Z-deformities may
arise at later stages. In general, JA is not characterized by
erosion or bone destruction in plain radography, unlike
rheumatoid arthritis. However, novel imaging modalities
such as magnetic resonance and high performance
ultrasonography revealed the presence of small erosions
in the joints of a few patients with JA. Pathophysiology
of JA still remains unknown. Deformities in JA are secondary to soft tissue abnormalities and include joints laxity, capsule fibrosis, synovitis tenosynovitis and muscular imbalance. Whether joint hypermobility contributes to development of JA is debatable. At present, JA treatment is conservative and is based on the use of non steroid anti inflammatory drugs, low dose corticosteroids, methotraxate, and antimarial drugs. Surgery may play role in treatment by realignment of soft tissue around joint or by more agressive procedures such as arthrodesis, silastic implant and arthroplasty. In cases with hallux valgus, osteotomy or resection can be carried out at metarsal head. In patients with metacarpophalangial joint fusion, joint functions may be improved with implant placement.

To date, coexistence of neurofibromatosis and Jaccoud arthropathy has not been reported. As mentioned before, this is the first reported case of association of Neurofibromatosis with JA. As other similar cases emerge, this issue will be elucidated further.
References


