Case report

Arthritis in Down's syndrome

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Summary. A 31 year old man with Down's syndrome presented with a 10 year history of an inflammatory polyarthritis resembling juvenile chronic arthritis. This case was similar to those already reported of an arthropathy associated with Down's syndrome but was eventually found to be gout. This emphasised the importance of serum uric acid estimation in patients with Down's syndrome and coexistent arthritis.

Key word: gout.

Musculoskeletal problems are well documented in Down's syndrome. Developmental anomalies including hypermobility are not uncommon and may lead to osteoarthritis.1 2 An arthropathy resembling juvenile chronic arthritis has been reported,3 but in the cases described treatment was not discussed and the outcome of the arthropathy was not stated. Our patient with Down's syndrome developed an arthropathy which was similar to those described.

Case report

A 31 year old man with classical Down's syndrome presented with a 10 year history of pains in large joints, particularly in the left elbow, knee, and ankle. He had also experienced some pain in the right elbow and knee. At first the arthritis came in attacks each lasting for about two weeks. One or more joints were involved and were often red, hot, and swollen. There was never any involvement of the small joints of the hands and feet. These attacks had become more frequent, and at the time of presentation he was rarely pain free. Although he had Down's syndrome, he had no other significant medical problems. A heart murmur had been noted at birth but had resolved spontaneously. Neonatal intestinal obstruction had been relieved surgically.

On examination at presentation he had a painful swollen right knee with a small effusion; it was not red. He had a warm swollen left ankle and some tenderness of both elbows, particularly the left, with normal flexion and extension but marked reduction in supination. There were no nodules, tophi, or other extra-articular manifestations. Investigations showed an erythrocyte sedimentation rate (ESR) of 86 mm/h (Westergren), haemoglobin of 111 g/l, white blood cell count 7·6×10⁹/l, rheumatoid factor (latex and RAHA) negative. Antinuclear antibodies were negative. An x ray examination of his joints showed no abnormalities in the hands or feet. The knees, however, showed narrowing of the medial compartment with radiolucent areas both in the femoral condyles and the tibial plateaux. In the elbows there was destruction of the radial head, with fusion to the medial border of the olecranon, and several radiolucent areas. There were no diagnostic features relating to rheumatoid arthritis, juvenile chronic arthritis (JCA), or gout.

Initially he made progress when treated with naproxen alone but continued to have painful exacerbations of his arthritis. He was treated with penicillamine but failed to respond; he was then treated with sulphasalazine with some improvement. A review of the case at this time prompted further investigation, which showed a serum uric acid of 0·64 mmol/l. Knee aspiration was performed at the next exacerbation of his arthritis and polarised light microscopy confirmed the presence of monosodium urate crystals. He was then treated with indomethacin, to which he responded instantly, and

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Table  Synopsis of clinical and laboratory features of the arthropathy of Down's syndrome

<table>
<thead>
<tr>
<th>Patient No</th>
<th>Sex</th>
<th>Age at presentation (years)</th>
<th>Joints involved</th>
<th>Systemic features</th>
<th>ANA†</th>
<th>Rheumatoid factor</th>
<th>ESR† (mm/h)</th>
<th>SUA† (mmol/l)</th>
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</table>

*Data for patients 1–7 taken from Yancey et al; patient No 8=our patient.
†ANA=antinuclear antibodies; ESR=erythrocyte sedimentation rate; SUA=serum uric acid.

his arthritis is now controlled by treatment with allopurinol. He has had no further attacks of arthritis.

Discussion

Down's syndrome is caused by trisomy of chromosome 21. It is associated with variable mental retardation and a series of congenital abnormalities.

Classic rheumatoid arthritis is rare, but a syndrome has been reported which resembles JCA. Seven cases of Down's syndrome with arthritis have been described (Table). Their age range at presentation was 7–22 years. Large joints were involved in an inflammatory arthropathy associated with negative antinuclear antibodies and rheumatoid factor. Unlike JCA, however, systemic features were absent.

Our patient had an inflammatory arthropathy beginning at the age of 20 with mainly large joint involvement, no systemic or extra-articular features, and negative tests for rheumatoid factor. He never had podagra. The x-ray appearances of affected joints showed no characteristic features of either rheumatoid arthritis, JCA, or gout. Although these features are all similar to those in cases described by Yancey et al, the patient was eventually found to have gout. Hyperuricaemia has frequently been reported in Down's syndrome, though only a few such cases develop gout. The metabolic abnormality causing this hyperuricaemia remains obscure. Nevertheless, the possibility of gout should be considered in a patient with Down's syndrome who develops arthritis. The diagnosis of gout is not always easy.

References

Arthritis in Down's syndrome.

J E Dacre and E C Huskisson

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