Down's syndrome

Objectives

1. List diagnostic features of Down's syndrome evident on physical exam
2. Describe features of the musculoskeletal system in children with Down's syndrome, including joint and bony abnormalities
3. Describe hip dysplasia associated with Down's syndrome
4. Describe the present data base for atlantoaxial instability in Down's syndrome
5. List lifethreatening non-orthopaedic problems associated with Down's syndrome

Discussion points

1. Why is the unstable hip so difficult to treat in a patient with Down's syndrome?
2. What joints other than the hip often cause difficulty for patients with Down's syndrome?
3. Is routine radiographic screening of the cervical spine necessary for children with Down's syndrome before participation in the Special Olympics?

Discussion

Down's syndrome results from a trisomy of the 21 chromosome, which has recently been virtually completely mapped (Hattori). Common musculoskeletal problems include atlantoaxial instability (which has been extensively studied), hip instability; and to a lesser degree, patellofemoral instability, flat feet, and bunions. Children with Down's syndrome exhibit generalized ligamentous laxity. A postmortem study on fetuses with Down's syndrome demonstrated grachcephaly, deficient ossification of the nasal bones, and hypoplasia of the middle phalanx of the little finger.

Non-orthopaedic problems include congenital heart disease (atrioventricular canal), bowel atresias, pulmonary hypoplasia, and vasculopathies of the basal ganglia. Almost 10% develop a 'transient leukemia’ during the first months of life, and there is a greatly increased incidence of acute myeloid leukemia in the first 4 years of life. Mortality of children with Down's is markedly affected by congenital heart disease.

The major orthopaedic problems are hip and upper cervical spine instability. The hip is difficult to treat, presumably from a combination of capsular laxity and bony dysplasia. Green has recently reported successful closed treatment in a small number of patients, although aggressive surgical management is usually recommended. Good results with total joint arthroplasty have been reported in adults with Down's syndrome.

The literature on atlantoaxial instability is huge and conflicting. In 1984, the American Academy of Pediatrics recommended routine radiographic examination before participation in the Special
Olympics, but reversed this recommendation in 1995, presumably as a result of papers similar to that by Ferguson, who found no difference in neurologic findings in children with Down's syndrome with increased C1-2 mobility compared with those who had a stable C1-2 articulation. This issue is presently unsettled. In addition, reports in the early 1990's detailed a high rate of serious complication following surgery to stabilize the C1-2 joint; more recent reports are somewhat more favorable, although the complication rate is still high. An unusually large bibliography accompanies this subject as the literature is so conflicted on the question of proper assessment and management of the upper cervical spine in the child with Down's syndrome.

References


