



# POZNA

## The Core Curriculum

### 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 rachicephaly, 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

1. Angelopoulou N, Tsimaras V, Christoulas K, Kokaridas D, Mandroukas K. Isokinetic knee muscle strength of individuals with mental retardation, a comparative study. *Perceptual & Motor Skills* 1999;88(3 Pt 1):849-55.
2. Aprin H, Zink WP, Hall JE. Management of dislocation of the hip in Down syndrome. *J Pediatr Orthop* 1985;5:428.
3. Brockmeyer D. Down syndrome and craniovertebral instability. Topic review and treatment recommendations. *Pediatric Neurosurgery* 1999;31(2):71-7.
4. Christensen JE, Peter PJ, Nielsen VK, Mai J. Prevalence of carpal tunnel syndrome among individuals with Down syndrome. *American Journal of Mental Retardation* 1998;102(6):547-51.
5. Diamond LSL, D., Sigman B. Orthopedic disorders in patients with Down's syndrome. *Orthop Clin North Am* 1981;12:57.
6. Dormans JP, Drummond DS, Sutton LN, Ecker ML, Kopacz KJ. Occipitocervical arthrodesis in children. A new technique and analysis of results. *Journal of Bone & Joint Surgery - American Volume* 1995;77(8):1234-40.
7. Doyle JS, Lauerman WC, Wood KB, Krause DR. Complications and long-term outcome of upper cervical spine arthrodesis in patients with Down syndrome. *Spine* 1996;21(10):1223-31.
8. Ferguson RL, Putney ME, Allen BL, Jr. Comparison of neurologic deficits with atlanto-dens intervals in patients with Down syndrome. *Journal of Spinal Disorders* 1997;10(3):246-52.
9. Frid C, Drott P, Lundell B, Rasmussen F, Anneren G. Mortality in Down's syndrome in relation to congenital malformations. *Journal of Intellectual Disability Research* 1999;43(Pt 3):234-41.
10. Greene WB. Closed treatment of hip dislocation in Down syndrome. *Journal of Pediatric Orthopedics* 1998;18(5):643-7.
11. Hasle H, Clemmensen IH, Mikkelsen M. Risks of leukaemia and solid tumours in individuals with Down's syndrome. *Lancet* 2000;355(9199):165-9.
12. Hattori M, Fujiyama A, Taylor TD, Watanabe H, Yada T, Park HS, et al. The DNA sequence of human chromosome 21. The chromosome 21 mapping and sequencing consortium [see comments]. *Nature* 2000;405(6784):311-9.
13. Higo M, Sakou T, Taketomi E, Kojyo T. Occipitocervical fusion by Luque loop rod instrumentation in Down syndrome. *Journal of Pediatric Orthopedics* 1995;15(4):539-42.

14. Kioschos M, Shaw ED, Beals RK. Total hip arthroplasty in patients with Down's syndrome. *Journal of Bone & Joint Surgery - British Volume* 1999;81(3):436-9.
15. Kriss VM. Down syndrome: imaging of multiorgan involvement. *Clinical Pediatrics* 1999;38(8):441-9.
16. Matsuda Y, Sano N, Watanabe S, Oki S, Shibata T. Atlanto-occipital hypermobility in subjects with Down's syndrome. *Spine* 1995;20(21):2283-6.
17. Pueschel SM, Scola FH, Tupper TB, Pezzullo JC. Skeletal anomalies of the upper cervical spine in children with Down syndrome. *Journal of Pediatric Orthopedics* 1990;10(5):607-11.
18. Pueschel SM, Scola FH, Pezzullo JC. A longitudinal study of atlanto-dens relationships in asymptomatic individuals with Down syndrome. *Pediatrics* 1992;89(6 Pt 2):1194-8.
19. Pueschel SM. Should children with Down syndrome be screened for atlantoaxial instability? [see comments]. *Archives of Pediatrics & Adolescent Medicine* 1998;152(2):123-5.
20. Rizzolo S, Lemos MJ, Mason DE. Posterior spinal arthrodesis for atlantoaxial instability in Down syndrome. *Journal of Pediatric Orthopedics* 1995;15(4):543-8.
21. Saad KF. A lethal case of atlantoaxial dislocation in a 56-year-old woman with Down's syndrome. *Journal of Intellectual Disability Research* 1995;39(Pt 5):447-9.
22. Segal LS, Drummond DS, Zanotti RM, Ecker ML, Mubarak SJ. Complications of posterior arthrodesis of the cervical spine in patients who have Down syndrome. *Journal of Bone & Joint Surgery - American Volume* 1991;73(10):1547-54.
23. Shikata J, Yamamuro T, Mikawa Y, Iida H, Kobori M. Atlanto-axial subluxation in Down's syndrome. *International Orthopaedics* 1989;13(3):187-92.
24. Stempfle N, Hutten Y, Fredouille C, Brisse H, Nessmann C. Skeletal abnormalities in fetuses with Down's syndrome: a radiographic post-mortem study. *Pediatric Radiology* 1999;29(9):682-8.
25. Taylor TK, Walter WL. Screening of children with Down syndrome for atlantoaxial (C1-2) instability: another contentious health question. *Medical Journal of Australia* 1996;165(8):448-50.
26. Toledo C, Alembik Y, Aguirre Jaime A, Stoll C. Growth curves of children with Down syndrome. *Annales de Genetique* 1999;42(2):81-90.
27. Tredwell SJ, Newman DE, Lockitch G. Instability of the upper cervical spine in Down syndrome. *Journal of Pediatric Orthopedics* 1990;10(5):602-6.
28. Uno K, Kataoka O, Shiba R. Occipitoatlantal and occipitoaxial hypermobility in Down syndrome. *Spine* 1996;21(12):1430-4.
29. York A, von Fraunhofer N, Turk J, Sedgwick P. Fragile-X syndrome, Down's syndrome and autism: awareness and knowledge amongst special educators. *Journal of Intellectual Disability Research* 1999;43(Pt 4):314-24.
30. Zipursky A, Brown E, Christensen H, Sutherland R, Doyle J. Leukemia and/or myeloproliferative syndrome in neonates with Down syndrome. *Seminars in Perinatology* 1997;21(1):97-101.