

**BASIC MEDICAL SURVEILLANCE ESSENTIALS
FOR PEOPLE WITH DOWN'S SYNDROME.**

THYROID DISORDER

(One of a set of guidelines drawn up by the Down's Syndrome Medical Interest Group)

1. At all ages thyroid disorder (usually hypothyroidism) occurs more frequently in people with Down's syndrome than in the general population^{1.2.3.4.5}. Around 10% of the school age population have uncompensated hypothyroidism. The prevalence increases with age⁶. If undiagnosed, thyroid disorder constitutes a significant cause of preventable secondary handicap. Diagnosis on clinical grounds is unreliable^{7.8}. Biochemical screening is essential. As in the general population those with significant abnormalities of any TFT should either be treated (if there is uncompensated hypothyroidism) or kept under close clinical and biochemical surveillance.
2. All babies in the U.K. have a neonatal screen for hypothyroidism⁹. For children with Down's syndrome each district should have a policy of screening after this, starting in infancy and continuing throughout life.
3. Biochemical testing, including estimation of T4, TSH, and thyroid antibodies should be carried out at least once every two years from age 1 and throughout life^{6.11}.
4. Fingerprick dried blood spot TSH measurement (Guthrie) is being investigated. Preliminary evaluation suggests that this may prove an effective screening procedure¹⁰. If available, and if replacing venous testing (see 3 above) this should be carried out at least annually.
5. Transient changes may occur.^{11.12} Mildly raised TSH (not greater than 10mu/l) or the presence of antibodies with normal T4 and no clinical evidence of hypothyroidism does not usually warrant treatment^{13.14}. It does however indicate increased likelihood of developing uncompensated hypothyroidism. Such people should therefore be tested more frequently than those with normal test results. A specialist opinion may be required.
6. Clinicians should always bear in mind the prevalence of thyroid disorder in people with Down's syndrome and have a low threshold for testing thyroid function if there is any clinical suspicion at times between biochemical testing.
7. As in the general population key clinical pointers are lethargy and/or changes in affect, cognition, growth, or weight.
8. Consideration of hypothyroidism is mandatory in the differential diagnosis of depression and dementia^{15.16}.
9. The possibility of hyperthyroidism should also be born in mind^{5.17}.

References (Thyroid disorder) :

1. Fort P, Lifshitz F, Bellisario R et al.(1984) *Abnormalities of thyroid function in infants with Down syndrome.* J Pediatr; **104** : 545-9.
2. Loudon MM, Day RE, Duke EMC (1985) *Thyroid dysfunction in Down's syndrome.* Archives of Disease in Childhood **60** : 1149-1151.
3. Sare Z, Ruvalcaba RHA, Kelley VC (1978) *Prevalence of thyroid disorders in Down syndrome.* Clin Genetics; **14**: 154-8.
4. Pueschel SM, Pezzullo JC (1985) *Thyroid dysfunction in Down Syndrome.* Am J Dis Child ; **139** : 636-9.
5. John JE, Cook AR (1962) *Hyperthyroidism in patients with Mongolism.* J Clin Endocrinol ; **22** : 665-8.
6. Prasher V (1995) *Reliability of diagnosing clinical hypothyroidism in adults with Down syndrome.* Aus. and NZ J of Developmental disabilities; **20**: 223 - 233.
7. Mani C (1988) *Hypothyroidism in Down syndrome.* Br J Psych; **153**: 102-4.
8. Quinn MW (1980) *Down's syndrome and hypothyroidism.* Ir J Med Sci; **149**: 19-22.
9. Grant DB, Smith I (1988) *Survey of neonatal screening for primary hypothyroidism in England, Wales and Northern Ireland 1982-84.* Br Med J; **296** : 1355-8.
10. Noble SE, Keyland K, Findlay CA, Clark CE, Redfern J, MacKenzie JM, Girdwood RWA, Donaldson MDC (2000) *School based screening for hypothyroidism in Down's syndrome by dried blood spot TSH measurement.* Arch.Dis.Chil. **82** 27-31
11. Selikowitz M (1993) *A five year longitudinal study of thyroid function in children with Down syndrome.* Dev. Med. Child Neurol; **35** ; 396-401.
12. Cutler AT, Benezra-Obeiter MD, Brink SJ (1986) *Thyroid function in young children with Down syndrome.* Am.J.Dis.Child. **140**479-483
13. Tirosh E, Taub Y, Scher A, Jaffe M, Hochberg Z (1989) *Short-term efficacy of thyroid hormone supplementation for patients with Down syndrome and low borderline thyroid function.* Am J of Mental Retardation ; **93** ; 652-6.
14. Vanderpump MPJ, Ahlquist JAO, Franklyn JA, Clayton RN on behalf of working group of RCP and Soc of Endocrinology. (1996) *Consensus statement for good practice and audit measures in the management of hypothyroidism and hyperthyroidism.* BMJ **313**. Aug 31st. 539-544
15. Thase ME (1982) *Reversible dementia in Down's syndrome.* J Ment Defic. Res ; **26** ; 111-3.
16. Collacott RA, Cooper S.A, McGrotherm C (1993) *Differential rates of psychiatric disorders in adults with Down's syndrome compared with other mentally handicapped adults.* Br J Psychiatry ; **161** : 671-74.
17. Takahashi H, Bordy MD, Sharma V, Grunt JA (1979) *Hyperthyroidism in patients with Down's syndrome.* Clinical paediatrics; **18** : 273 - 275.

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