

MULTIPLE ENDOCRINE CHANGES DEVELOPING IN A MAN WITH DOWN'S SYNDROME: A Case Report

R. Laugharne and E. Akuffo

We report the case of a man (P.T.) with Down's Syndrome who has been presented with endocrine changes in adult life. Thyroid abnormalities have been known to be associated with Down's Syndrome for some years (Korsager *et al.*, 1978), but there is no reported association between abnormalities in pituitary function and Down's Syndrome.

Case Report

The patient is a 32 year old caucasian man with Down's Syndrome. His mother was 42 years old when he was born and he is living with her at present. His father, who died at the age of 72 years from carcinoma of the lung, had a career in the army and was described as

large in stature. The patient has one brother and four sisters and relations in the family are said to be good. There is no family history of endocrine abnormality or psychiatric illness.

In spite of his Down's Syndrome early developmental milestones were described as apparently normal and his learning disability is of a moderate degree. He has lived at home without needing a great deal of support, having been cared for and having companionship from his family. He currently spends much of his time with his mother and is able to function well in his activities of daily living. Occasionally he will get frustrated by difficulties in finding the right words but he is a happy and placid man, attending a local centre for sheltered work weekly and another day centre for people with learning dis-

* **Richard Laugharne, BM BS, MRC Psych.**

Registrar in Psychiatry

Department of Psychiatry, Princess Alexandra Hospital, Hamstel Road, Harlow, Essex
CM20 1QX

Emmanuel Akuffo, FRCPsych. DPH

Senior Lecturer in Psychiatry of Learning Disabilities
St. Bartholomew's Hospital Medical School, London

* *For Correspondence*

abilities regularly. Attendance at other activity centres and art therapy has stopped because he has not wished to continue. His mother is protective towards him and tends to encourage him to be dependent on her, but at times she seems to be dependent on his companionship as well.

He is on no medication and there are no features of formal psychiatric illness.

P.T. has never needed to shave throughout his adult life but in the past had pubic hair. Five years ago his pubic hair began to disappear and his penis appeared to diminish in size. At present only the end of the glans penis and his urethra can be seen, whilst his testes are palpable. Over the last year he has increased in weight from 76 kilograms to 114 kilograms and developed a maculopapular rash on his lower abdomen.

The only past medical history was the need for circumcision when he was ten years old for phimosis. Investigations revealed raised TSH, prolactin, FSH and ESR but normal T4, 24 hour urine cortisol and LH.

Liver function tests showed raised alkaline phosphatase and gamma-globulin but other values were normal. There were no other significant findings on blood examination and a lateral skull x-ray showed no abnormality of the pituitary fossa.

Discussion

This report of a man with Down's Syndrome developing abnormal endocrine functioning in adult life can be

taken in the context of a known association between Down's Syndrome and thyroid abnormality (Korsager *et al.*, 1978, Mani, 1988). Pozzan *et al.* (1990) in a sample of 108 patients with Down's Syndrome found five hypothyroid, two hyperthyroid and thirty three had high TSH values with T4 levels in the normal range, indicating a high level of subclinical hypothyroidism. In the same study antithyroid autoantibodies were found in thirteen patients. Zori *et al.* (1990) found in 61 patients with Down's Syndrome that 35 had abnormal TSH levels, 3 Hashimoto's thyroiditis and 2 Graves' disease. They found 48% of those with thyroid dysfunction over 10 years of age had thyroid autoantibodies compared with 20% of those under 10 years with thyroid abnormality. Of other autoantibodies, one patient had gastric parietal cell autoantibodies and no patients had antibodies to the adrenal cortex. They suggested that thyroid abnormality in Down's Syndrome was a common heterogeneous disorder which cannot be solely explained on the basis of autoimmunity.

Napolitano *et al.* (1990) have suggested that the finding of early thymic involution in patients with Down's Syndrome may be associated with pituitary-thyroid axis dysfunction. In a study of 52 patients they showed an improvement in thyroid function with zinc supplementation in nine patients with low zinc levels: zinc supplementation has also been reported to transiently improve thymic function.

Percy *et al.* (1990) in a sample of twenty nine patients with Down's Syn-

drome found in a case control study that subclinical hypothyroidism was more prevalent in those with Alzheimer's disease than those without, suggesting that subclinical hypothyroidism could be related to cognitive deficits.

Summary

Whilst the above studies have made reference to an association between the pituitary-thyroid axis and Down's Syndrome, most have focused on thyroid abnormality and autoantibodies in those with Down's Syndrome. No adrenocortical autoantibodies were found in a sample of 61 persons with Down's Syndrome (Zori *et al.*, 1990) but a possible link between thymic and thyroid functions has been suggested (Napolitano *et al.*, 1990). The case report above may show purely a coincidental concurrence of multiple endocrine abnormalities and Down's Syndrome, but we suggest further studies investigating a link between pituitary function and Down's Syndrome may be indicated.

References

- Korsager, S., Chatham, E. M. and Ostergaard, M. P. (1978). Thyroid function tests in adults with Down's Syndrome. *Acta Endocrinologica*, 88, 48-54.
- Mani, C. (1988). Hypothyroidism in Down's Syndrome. *British Journal of Psychiatry*, 153, 102-104.
- Napolitano, G., Palka, G., Lio, S., Bucci, I., De Remigis, P., Stuppia, L. and Monaco, F. (1990). Is zinc deficiency a cause of subclinical hypothyroidism in Down's Syndrome? *Annales De Genetique*, 33(1), 9-15.
- Percy, M. E., Dalton, A. J., Markovic, V. D., Crapper McLachlan, D. R., Gera, E., Hummel, J. T., Rusk, A. C., Somerville, M. J., Andrews, D. F. and Wagish, P. G. (1990). Autoimmune thyroiditis associated with mild subclinical hypothyroidism in adults with Down's Syndrome: a comparison of patients with and without manifestations of Alzheimer disease. *American Journal of Medical Genetics*: 36(2), 148-154.
- Pozzan, G. B., Rigon, F., Girelli, M. E., Rubello, D., Busnardo, B. and Bassicheti, C. (1990). Thyroid Function in patients with Down's Syndrome. *American Journal of Medical Genetics: Supplement* 7, 57-58.
- Zori, R. T., Schatz, D. A., Ostrer, M., Williams, C. A., Spillar, R. and Riley W. J. (1990). Relationship of autoimmunity to thyroid dysfunction in children and adults with Down's Syndrome. *American Journal of Medical Genetics: Supplement* 7, 238-241.