

Success Stories
in
Developmental Disabilities

Volume II
Dale Antanitus, M.D., Editor

Presented in part at the AAMR 117th Annual
Meeting - Washington, D.C.
June 3, 1993

HYPOTHYROIDISM PRESENTING AS SEVERE PSYCHOLOGICAL AND
MENTAL DYSFUNCTION IN AN ADULT WITH DOWN SYNDROME

Brian Chicoine, M.D.

Hypothyroidism is seen more frequently in Down Syndrome (DS) than in the general population (1,2). Dry skin, constipation, impaired intellect, obesity, hearing impairment, lethargy and depression are manifestations of hypothyroidism(3). They can also be manifestations of DS or associated health problems. Therefore, it can be difficult to determine if an adult with DS has hypothyroidism using clinical findings alone(4).

The pathologic changes of the brain seen in Alzheimer's Disease(AD) are also seen more frequently in persons with (DS) (5). Numerous patients have been seen at our clinic with a presenting complaint that the adult with DS has deteriorating capabilities. Often the family or care giver is concerned about the possibility of Alzheimer's disease or has previously been told that the patient has Alzheimer's disease.

F. is a 32 year old male whom I met at his home because he had not been out the house for more than eight years. His mother noted that his activity level had been slowing down for about 20 years. In addition, since his father died one and a half years prior to my visit, he would not get out of bed, he refused to eat for anyone except his mother, required

spoon feeding, had become regularly incontinent of urine and stool, was up frequently during the night, frequently screamed and appeared "depressed" to his mother. In addition, he was overweight, had dry skin, and was weak and lethargic. He had poor verbal skills making assessment of his emotional state difficult through interview. However, the Checklist for Psychological Concerns was used which supported that clinical impression of a major depressive episode.

CBC, SMAC, folate, B-12, RPR, urinalysis, stool cultures and ova and parasites and ESR were all normal. However, his TSH was 12.6 (normal: 0.32-5.0 uIU/ml) while his T3 was 43.8 (30-40%), and T4 was 4.6 (4.5-12.0 ug/dl). He was started on Synthroid 0.025mg daily and this was gradually adjusted as guided by laboratory values. His TSH normalized with Synthroid 0.075mg daily. The only additional therapy was that a respite worker started going to the house for four hours twice a week.

The second visit to his house four months after the first revealed a dramatic change. He was now getting out of bed, was using the toilet regularly and was rarely incontinent, was eating a variety of foods, was attending to more of his own daily care, was no longer screaming randomly and was sleeping through the night. His mother felt he was much happier and his mother appeared much more relaxed.

In addition to the obvious improvement in the health of F., his mother was able to get to her own physician and have the mammogram which she had been putting off because of her inability to leave the home. A small, non-palpable mass was found. The malignancy was able to be removed long before it ever would have been apparent to her.

Discussion:

Hypothyroidism is a common problem in DS with incidence reported from 2% to 63% (2). The incidence of depression in adults with DS is less well studied but ranges from 6% to 14% (6,7) Hypothyroidism is one cause of depression.

Depression can be a very debilitating illness. DSM-III criteria include dysphoric mood, poor appetite, sleep disturbance, psychomotor agitation or retardation, loss of interest, loss of energy, feelings of worthlessness, diminished ability to think or concentrate and/or thoughts of death or suicide(8). In persons with DS, the symptoms may differ from the general population and may include antisocial behavior, repetitive/stereotypic movements, temper tantrums, and screaming/crying episodes(9).

Much concern has been raised with regards to the development of Alzheimer's disease in adults with DS. While autopsy findings may find pathologic changes consistent with AD, the functional changes seen in adults with DS can be a manifestation of a multitude of other medical or psychiatric problems as well(9). A major difference is the reversible nature of many of these other illnesses.

While AD is a concern for adults with DS, the presentation of decreased functioning in adults with DS can have many etiologies including depression, hypothyroidism, and others. The case of F. illustrated a patient who was felt by his family and the referring social agency to have AD. Through evaluation he was found to have depression and hypothyroidism. While etiology of his depression is probably multifactorial and will continue to require further intervention, a major change in his life and the life of his mother was realized with the treatment of his hypothyroidism. While further research is needed in the area of AD in general and in DS, specifically, each patient must be evaluated to search for potential reversible causes of dementia or dementia-like reduction in function.

References

1. Pueschel, SG., Jackson, IMD., Giesswein, P., Dean, MK., & Pezzullo, JC. (1991) Thyroid Function in Down Syndrome. Research in Developmental Disabilities. 12: 287-296.
2. Friedman, DL., Kastner, T., Pond, WS., & O'Brien, DR. (1989) Thyroid Dysfunction in Individuals with Down Syndrome. Arch Intern Med. 149:1990-1993. 1993.
3. Wyngaarden, JB. & Smith, LH. (eds.) (1988). Cecil Textbook of Medicine. WB Saunders Co. Philadelphia, pp. 1328-1331.
4. Pueschel, SM & Pueschel, JK. (eds) (1992). Biomedical Concerns in Persons with Down Syndrome. Paul H Brookes Publishing Co. Baltimore, p. 266.
5. Wisniewski, KE., Wisniewski, HM., Wen, GY. (1985). Occurrence of Neuropathologic Changes and Dementia of Alzheimer's Disease in Down's Syndrome. Ann Neurol 17: 278-282
6. Meyers, BA., & Pueschel, SM. (1991). Psychiatric Disorders in Persons with Downs Syndrome J Nerve Ment Dis 179: 609-613.
7. Collacott, RA., Cooper, SA., & McGrother C. Differential Rates of Psychiatric Disorders in Adults with Down's Syndrome Compared with Other Mentally Handicapped Adults. British Journal of Psychiatry 161: 671-674.
8. American Psychiatric Association (1980). Quick Reference to the Diagnostic Criteria from Diagnostic and Statistical Manual of Mental Disorders. Third Edition. Washington, DC. pp 120-122. Washington, DC. pp. 120-122.

9. Pueschel SM & Pueschel JK. (eds) (1992) Biomedical Concerns in Persons with Down Syndrome. Paul H. Brooks Publishing Co. Baltimore. pp. 186-187.

Editor's Comment: A doctor who still makes house calls!