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## Clinical Info: Endocrine Conditions in Down Syndrome

By: Dr. Ernest McCoy

Individuals with Down syndrome have a higher incidence of endocrine problems than the general population. The endocrine system refers to a set of glands which includes the thyroid, adrenal and pituitary glands.

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### What is hyperthyroidism?

In this case, the thyroid gland is overactive. Symptoms are swelling in the neck, abnormal sweating and rapid pulse rate. No evidence exists as to whether hyperthyroidism is more prevalent in individuals with Down syndrome than in the general population.

### What is hypothyroidism?

hypothyroidism results from a malfunctioning thyroid gland, which decreases the synthesis of the hormone thyroxin. Thyroxin is the hormone that promotes growth of the brain and other body tissue.

### How common is hypothyroidism?

Hypothyroidism is the most common endocrine problem in

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children with Down syndrome. One study showed these individuals to be 28 times more prone to congenital hypothyroidism than the general population. It is estimated that approximately 10 percent of children with Down syndrome have congenital or acquired thyroid disease. Incidence of thyroid disease in adults with this genetic disorder varies from 13 to 50 percent. Hypothyroidism can occur at any time from infancy through adulthood.

### **How is hypothyroidism diagnosed?**

All individuals with Down syndrome should be tested for hypothyroidism at birth and at periodic intervals (at least every two years) thereafter. The indicators of hypothyroidism – enlarged tongue, constipation, poor circulation – are also found in individuals who are not hypothyroid, so the blood test for thyroid function is an important diagnostic test. As the thyroid hormone affects normal development of the brain, testing of infants is particularly crucial.

### **What treatment exists for hypothyroidism?**

The thyroid hormone, thyroxin, is readily replaced through medication.

### **Are people with Down syndrome more prone to diabetes?**

There is not sufficient data available at this point to know if there is increased risk for children with Down syndrome to develop type one diabetes as compared to the rate for their peers in the general population. However, research suggests that individuals who develop one type of endocrine autoimmune disorder, such as thyroiditis, are more likely to develop a second disorder, such as type one diabetes.

### **What is the status of research on use of the growth hormone for children with Down syndrome?**

Use of growth hormone for children with Down syndrome is still in experimental stages. There are reports of increased rates of growth in children with Down syndrome who received the hormone for a brief period; however, these reports were not controlled studies, so there is no scientific evidence that long-term administration of the hormone would increase final height.

### **Can any growth hormone be administered to any Down syndrome child?**

At present, doctors are licensed to prescribe growth hormone for individuals with Down syndrome only when there is a demonstrated deficiency of that hormone. It is strongly advised that such treatment be obtained only through a clinical trial until long-term benefits are demonstrated.

**Summary:**

- Approximately 10 percent of children and between 13 to 50 percent of adults with Down syndrome have thyroid disease. Hypothyroidism, an under-functioning thyroid gland, is the most common condition.
- Hypothyroidism must be diagnosed by a standard blood test, beginning at infancy and continuing at regular intervals throughout the life span.
- Hypothyroidism is easily treated with replacement thyroxine hormone; treatment is particularly crucial for hypothyroid infants, as the hormone affects normal development of the brain. However, this thyroid disease can be developed at any time in the life span.
- Hyperthyroidism is an overactive thyroid gland. Diagnosis is also by blood test. The condition can be treated by medication, surgery or use of radioactive iodine to destroy the gland.
- Insufficient evidence exists on the issue of whether individuals with Down syndrome are more prone to type one diabetes. Research does suggest that individuals who develop one type of endocrine autoimmune disorder, such as thyroid disease, run a higher risk of developing other autoimmune disorders, such as type one diabetes.
- While some short-term uncontrolled studies on the use of growth hormone for children with down syndrome have demonstrated growth spurts, there is no long-term research to demonstrate an increase in final height. Growth hormone should be administered only as part of a clinical trial until long-term benefits are demonstrated.
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**Books/Newsletters:**

Endocrine Conditions in Down Syndrome, McCoy, E. (1995). New York, NY: National Down Syndrome Society. Available through the National Down Syndrome Society, 666 Broadway, New York, NY 10012; tel.: (800) 221-4602 or (212) 460-9330.

This booklet provides general information about the thyroid, adrenal and pituitary glands of the endocrine system and answers commonly asked questions about medical conditions of the endocrine system, as they relate specifically to Down syndrome.

"Endocrine Function in Down Syndrome," McCoy, E. In: Down Syndrome: Advances in Medical Care, Lott, I. & McCoy, E. (Eds.) (1992). New York, NY: Wiley-Liss, pp. 71-82. Available through Wiley-Liss, 1 Wiley Dr., Somerset, NJ 08875; tel.: (800) 225- 5945.

This chapter provides information about three areas of endocrine function in individuals with Down syndrome, including thyroid function, gonadal function and growth

patterns. The chapter is written in technical language.

"Endocrinologic Aspects," Pueschel, S. & Bier, J. In: Biomedical Concerns in Persons with Down Syndrome, Pueschel, S. & Pueschel, J. (Eds.) (1992). Baltimore, MD: Paul H. Brookes, pp. 259-272. Available through Paul H. Brookes Publishing Co., P.O. Box 10624, Baltimore, MD 21285-0624; tel.: (800) 638-3775.

This chapter provides an overview of the various conditions associated with the endocrine system and examines the functioning of the hormones and glands, specifically in individuals with Down syndrome.

"The Endocrine System" In: Medical Care in Down Syndrome: A Preventive Medicine Approach. Rogers, P. & Coleman, M. (1992). New York: NY Marcel Dekker, Inc., pp.189-200. Available through Marcel Dekker, Inc., 270 Madison Ave., 4th Floor, New York, NY 10016; tel.: (212) 696-9000.

This chapter discusses abnormalities of the endocrine system, relating specifically to thyroid and gonadal functions. The authors stress the importance of routine thyroid screenings.

"Thyroid Conditions and Other Endocrine Concerns in Children with Down Syndrome," Foley, T., Jr. In: Medical & Surgical Care for Children with Down Syndrome: A Guide for Parents. Van Dyke, D., Mattheis, P., Eberly, S. & Williams, J. (Eds.) (1995). Bethesda, MD: Woodbine House, pp. 85-108. Available through Woodbine House, 6510 Bells Mill Rd., Bethesda, MD 20817; tel.: (800) 843-7323.

This chapter provides a general overview of the endocrine system and the various related medical concerns, including thyroid disease and diabetes mellitus. The authors also address symptoms, diagnosis and treatments for endocrine-related problems. The text is written in simple language.

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