



## HYPERTHYROIDISM

# There Is An Increased Incidence Of Hyperthyroidism In Down Syndrome That Should Be Regularly Tested For At All Ages

### WHAT IS THE STUDY ABOUT?

Down syndrome is the most common genetic form of mental retardation and the leading cause of certain birth defects and medical conditions. Remarkably, Down syndrome patients share many similarities in appearance and in medical conditions regardless of their racial or ethnic background. Thyroid disorders are common in patients with Down syndrome, with hypothyroidism occurring in almost 50%. There have been a few cases reported of hyperthyroidism in these patients. It is unknown if hyperthyroidism is seen more often in patients with Down syndrome. The aim of this study was to determine how common hyperthyroidism was in a population with Down syndrome and how these patients were diagnosed and treated.

### THE FULL ARTICLE TITLE:

Goday-Arno A, Cerda-Esteva M, Flores-Le-Roux JA, Chillaron-Jordan JJ, Corretger JM, Cano-Perez JF. Hyperthyroidism in a population with Down syndrome (DS). Clin Endocrinol (Oxf) 2009;71:110-4.

### WHAT WAS THE AIM OF THE STUDY?

The aim of this study was to determine how common hyperthyroidism was in a population with Down syndrome and how these patients were diagnosed and treated.

### WHO WAS STUDIED?

The study group included 1832 patients registered with the Catalan Down Syndrome Foundation in Spain. The information on these patients was obtained from January 1991 through February 2006.

### HOW WAS THE STUDY DONE?

The records of 1832 patients registered with the Catalan Down Syndrome Foundation in Spain were reviewed. The following information was gathered: age, vital signs, symptoms and physical examination, thyroid blood test results, thyroid scan results, ultrasound result and diagnosis.

### WHAT WERE THE RESULTS OF THE STUDY?

Among 1832 individuals with Down syndrome, 12 had hyperthyroidism, 5 of whom were male and 7 of whom were female, with ages ranging from 10.9 to 28.9 years.

The incidence of Graves' disease in the Down syndrome population was 43 cases per 10,000 persons per year, as compared with 24 cases per 100,000 persons per year in the population of Spain. The most common symptoms leading to a diagnosis of hyperthyroidism were increased heat intolerance and sweating (92%), increased emotional irritability (83%), a mean weight loss of 25.1 lb, palpitations (75%), insomnia (58%) and shaking of the hands (58%). The thyroid examination revealed diffuse thyroid enlargement in all the patients. All patients had undetectable serum TSH concentrations with elevated serum FT<sub>4</sub> concentrations and elevated serum total T<sub>3</sub> concentrations. Thyroid antibodies were present in 11 of 12 patients (92%). A thyroid scan showed diffuse thyroid uptake in all patients, which was suggestive of Graves' disease. All patients were initially treated with antithyroid drugs and 10 (83%) eventually were treated with radioactive iodine which resulted in hypothyroidism requiring levothyroxine therapy in all patients.

### HOW DOES THIS COMPARE WITH OTHER STUDIES?

The association of Down syndrome with thyroid disorders has been recognized for decades and up to 54% of individuals with Down syndrome are reported to have thyroid disorders, mainly hypothyroidism. The recommendation is to regularly screen individuals with Down syndrome for hypothyroidism. A total of 46 cases of hyperthyroidism have been previously reported in Down syndrome but details of the therapy were unclear in most of these cases.

### WHAT ARE THE IMPLICATIONS OF THIS STUDY?

There is an increased incidence of hyperthyroidism, specifically Graves' disease, in Down syndrome. Most patients have typical symptoms of hyperthyroidism. Physicians taking care of these patients need to be aware of this association and consider a low threshold for obtaining thyroid function tests.

— Alan Farwell, MD

### ATA THYROID BROCHURE LINKS

Hyperthyroidism: [http://thyroid.org/patients/patient\\_brochures/hyperthyroidism.html](http://thyroid.org/patients/patient_brochures/hyperthyroidism.html)

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## **HYPERTHYROIDISM**, continued

### **ABBREVIATIONS & DEFINITIONS**

**Down syndrome:** the most common genetic form of mental retardation and the leading cause of certain birth defects and medical conditions.

**Hyperthyroidism:** a condition where the thyroid gland is overactive and produces too much thyroid hormone. Hyperthyroidism may be treated with antithyroid meds (Methimazole, Propylthiouracil),

radioactive iodine or surgery.

**Graves' disease:** the most common cause of hyperthyroidism in the United States.

**Methimazole:** an antithyroid medication that blocks the thyroid from making thyroid hormone. Methimazole is used to treat hyperthyroidism, especially when it is caused by Graves' disease.