



Case 15.1 Graves' disease

A 29-year-old woman presented with a 3-month history of increased sweating and palpitations with weight loss of 7 kg. On examination, she was a nervous, agitated woman with an obvious, diffuse, non-tender, smooth enlargement of her thyroid, over which a bruit could be heard. She had a fine tremor of her fingers and a resting pulse rate of 150/min. She had no evidence of exophthalmos. A maternal aunt had suffered from 'thyroid disease'.

On investigation, she had a raised serum T3 of 4.8 nmol/l (NR 0.8–2.4) and a T4 of 48 nmol/l (NR 9–23). Measurement of her thyroid-stimulating hormone showed that this was low normal, 0.4 mU/l (NR 0.4–5 mU/l). The biochemical findings pointed to primary thyroid disease rather than pituitary overactivity. Circulating antibodies to thyroid peroxidase (titre 1/3000; 200 IU/ml) were detected by agglutination. A diagnosis of autoimmune thyrotoxicosis (Graves' disease) was made. She was treated with an antithyroid drug, carbimazole, to control her thyrotoxicosis, and surgery was not required.



Case Figure 15.1 Graves' disease – Case Figure states no exophthalmos but picture shows the thyrotoxic mother with exophthalmos and the infant with transient thyroid hyperactivity which resolves without treatment as the maternal IgG (transferred via the placenta) is metabolised over 3–4 months. Roitt & Rabson 2000.



Case 15.2 Hashimoto's thyroiditis

A 39-year-old woman presented with a large, painless swelling in her neck. The enlargement had been a gradual process over 2 years. She had no other symptoms and felt generally well. On examination, her thyroid was diffusely enlarged and had a rubbery consistency. There were no signs of thyrotoxicosis or of thyroid failure on clinical examination.

Thyroid function tests showed that she was euthyroid; T3 was 1.2 nmol/l (NR 0.8–2.4), T4 was 12 nmol/l (NR 9–23) and TSH was 6.3 mU/l (NR 0.4–5 mU/l). However, her serum contained high-titre antibodies to thyroid peroxidase (1/64 000; 4000 IU/ml).

This patient had Hashimoto's thyroiditis. The goitre was huge, and she was treated by partial thyroidectomy; the goitre did not recur, and the patient has remained euthyroid for 12 years.



Case 15.3 Primary myxoedema

A 41-year-old woman complained to her doctor that she 'always felt cold', and that she had become increasingly clumsy. Although she made no other complaint, her husband had noticed increasing physical and mental lethargy in his wife in recent months. One of her sisters had thyroid disease and her mother suffered from pernicious anaemia. On examination, her skin was dry, her voice was hoarse and her hair was coarse and brittle. Her pulse rate was 58/min, with a blood pressure of 140/70. Her tendon reflexes showed a markedly delayed relaxation phase.

Clinically, she had hypothyroidism and this was confirmed by thyroid function tests; her serum T3 was 0.4 nmol/l (NR 0.8–2.4), T4 was 4 nmol/l (NR 9–23), and TSH was 12.1 mU/l (NR 0.4–5 mU/l). High titres of autoantibodies to thyroid peroxidase were found in the patient's serum to a titre of 1/128 000 (6400 IU/ml). This patient therefore had primary myxoedema and she was treated with replacement doses of L-thyroxine.



Case 15.4 Diabetes mellitus

A 26-year-old pregnant woman attended the antenatal clinic regularly. She had no family history of diabetes. At 24 weeks' gestation she was found to have asymptomatic glycosuria. A glucose tolerance test showed that not only was her fasting blood glucose raised but that she had poor glucose tolerance. Gestational diabetes was diagnosed and the patient was admitted for diabetic control. This was achieved on oral hypoglycaemic agents alone and the patient was instructed to check her urine daily. The pregnancy was uneventful and a normal, 3.8 kg baby was born. The patient's glucose tolerance returned to normal in the puerperium; however, her serum, which was found to contain antibodies to pancreatic islet cells at the time of diagnosis, remained positive. Nine years later, after yearly checks, the patient developed overt diabetes mellitus.



Case 15.5 Addison's disease

A 12-year-old girl presented with vague abdominal discomfort for 6 months. She had noticed occasional diarrhoea but had not passed any blood. She admitted to weight loss (6 kg) and anorexia. On examination, she was obviously pigmented, although she thought this was sun induced; however, her buccal mucosa and gums were also brown. There were no other physical signs.

She had a low cortisol level and her response to the adrenocorticotrophic hormone in a Synacthen test was poor. A diagnosis of adrenal cortical failure was made. X-ray of her abdomen showed no calcified areas in either adrenal gland, and her serum contained antibodies to adrenal cortex, consistent with a diagnosis of Addison's disease due to autoimmune adrenalitis. Her serum also contained antibodies to pancreatic islet cells and thyroid microsomes. In view of her young age at presentation and these serum antibodies, she will be followed at yearly intervals to see if she develops other autoimmune endocrinopathies.