Case 5.7 Primary autoimmune hypothyroidism

A 36-year-old woman presented with a 1-year history of 20-kg weight gain, generalized fatigue, thinning hair and tingling in her hands at night. Her husband had also commented that she had developed a deeper, croaky voice. Her periods had become irregular and lighter than previously. On examination she was overweight with generally dry skin and thin, rather coarse hair. She was bradycardic with a pulse of 60. She had evidence of bilateral carpal tunnel syndrome and was felt to have slow relaxing reflexes. Investigation confirmed the clinical diagnosis of primary hypothyroidism, her serum thyroid-stimulating hormone (TSH) was significantly elevated at 28 U/ml (normal <4) and her total serum thyroxine was low at 19 μmol/l. Antibodies to thyroid peroxidase were present at high titre, consistent with primary autoimmune hypothyroidism.

She was treated with oral thyroxine at 100 μg daily. Her TSH and serum thyroxine returned to the normal range and her skin, hair, voice and hands returned slowly to normal over the next year. Her weight however remained 15 kg heavier than before she developed myxoedema.

She agreed to take part in a family study of autoimmune disease. The prevalence of autoantibodies and autoimmune disease are summarized in Case Figure 5.7.

Case Figure 5.7 Family study of Case 5.7. Males, square symbols; females, round symbols; Thy AB+, serum contains antibodies to thyroid peroxidase; Myxo, myxoedema.
Case 5.8 IL-2 treatment and rheumatoid arthritis

A 53-year-old man presented to his GP with pain in the right loin and intermittent haematuria. His only medical history was of mild rheumatoid arthritis which had presented at the age of 42 but which was currently in remission without any additional medication.

Examination revealed a large mass apparently arising from the right kidney. Subsequent investigation by a urologist demonstrated a large tumour replacing most of the right kidney but with no evidence of distant metastasis. He underwent a right nephrectomy. Histological examination confirmed that the tumour was a renal cell carcinoma.

He remained well for the next 2 years but then developed severe back pain. Further radiological studies demonstrated widespread metastases in bone, liver and lungs. He underwent treatment with recombinant human IL-2 as part of a clinical trial of combined chemotherapy and immunotherapy. Ten days after receiving IL-2, he developed a severe flare of his rheumatoid arthritis, necessitating treatment with corticosteroids. No further IL-2 was administered and his arthritis settled. His disseminated malignant disease did not respond to chemotherapy and he died 8 weeks later in his local hospice.