An extreme case of neurofibromatosis type 1

Neurofibromatosis type 1 or von Recklinghausen disease is one of the most common autosomal dominant genetic disease and is associated with plexiform neurofibromata [1]. They affect peripheral nerves, leading occasionally to a massive overgrowth of nerves and connective tissue [2, 3]. A 59-year-old man with neurofibromatosis type 1 was admitted to our clinic because of tumour mass overgrowth (weight 170 kg; circumference of the tumour mass 190 cm), scrotum oedema, dyspnoea and oliguria. He was unable to walk; large subcutaneous neurofibromata with *café-au-lait* spots were palpable over both gluteal regions and thighs, he had scrotal pain. Urine volume was less than 500 ml/day. Soon after his birth, *café-au-lait* spots had been noticed and painful subcutaneous nodules had appeared. In adolescence tissue on the back, gluteal region, lower abdomen and upper parts of the legs had grown considerably. At age 57 he had had a rupture of the right upper leg which had been treated surgically. Since then the tumour mass had grown extensively. On admission urea was 29.4 mmol/L, creatinine 294 mmol/L, potassium 5.0 mmol/L, sodium 126 mmol/L, Hgb 82 g/L. Abdominal and pelvic sonography showed highly vascularised giant peduncular neurinomas. He was treated with furosemid 250 mg daily and ACE inhibitors. He lost more than 35 L of fluids in two weeks. Gradually, scrotal oedema diminished, the tumour mass was moderately reduced, urea and creatinine values normalised and he was able to perform daily activities.

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References

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