TITLE

VON HIPPEL-LINDAU SYNDROME WITH MUSCULOSKELETAL MANIFESTATIONS. REPORT OF A CASE

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OBJECTIVES

Von Hippel-Lindau (VHL) syndrome is an inherited disorder characterized by the formation of tumors and cysts in diverse organ systems. Tumors may be malignant or benign and they tend to appear during young adulthood. Hemangioblastomas are characteristic of VHL syndrome.

The aim was to describe the case of a patient with VHL syndrome presenting with musculoskeletal manifestations and multiple tumors.

METHODS

A patient female, aged 51 years, presented with painful 1st and 3rd MCP and proximal PP joints. The patient complained of dry mouth and dry eyes. Laboratory investigations revealed positive RF test. The patient had a 10-year history of insulin treated diabetes mellitus type 2 on insulin and antidiabetic drugs. At the age of 27 the patient developed loss of balance, headaches and vision problems. An MRI performed revealed a hemangioblastoma of the cerebellum. The diagnosis of VHL syndrome was made.

Graphs and tables

RESULTS

At the age of 49 she presented with multiple hemangioblastomas of the cerebellum and the spinal cord. There were multiple cases of VHL syndrome within the family. Her mother was diagnosed with VHL at the age of 57, had multiple hemangioblastomas in the CNS and died from clear cell renal carcinoma aged 73. Her sister has also multiple hemangioblastomas and has been diagnosed with clear cell renal carcinoma and has also VHL. Her first son was diagnosed with VHL at the age of 12, developed multiple hemangioblastomas in the CNS causing tetraplegia and died aged 25. Her second son has multiple hemangioblastomas in the cerebellum, a pancreatic serous cystadenoma and had a retinal angioma in the right eye with subsequent loss of vision. Her 2 nephews also suffer from multiple hemangioblastomas in the CNS, one of them having a retinal angioma.

CONCLUSIONS

The extremely rare case of a patient with VHL syndrome and diabetes mellitus type 2 is described. The syndrome is inherited in an autosomal dominant pattern, thus patients present with multiple cases within the family.

References

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