Case Report Recurrent Cerebellar Hemangioblastoma in Two Siblings Carrying Von Hippel Lindau Syndrome

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Introduction

Von Hippel-Lindau Syndrome is an autosomal dominant genetic disease caused by mutations in the VHL gene (1).

The presence of cerebellar hemangioblastomas keep being the main cause of mortality in these patients (2).

This is a case repot of two siblings presenting recurrent cerebellar hemangioblastoma carrying VHL syndrome.

Methods

This is an observatory descriptive case report study

Objective

To present two cases of cerebellar hemangioblastomas in two siblings, who received surgical treatment as first line treatment and currently underwent SRS in October 2024.

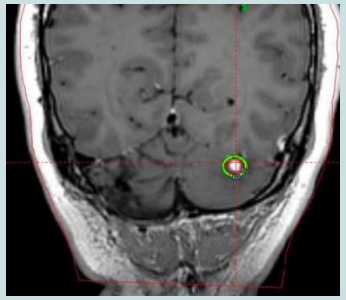
Case report

Two family cases of cerebellar hemangioblastomas are presented in siblings from Lima, Peru. Both patients were carriers of Von Hippel-Lindau Syndrome. The initial approach for both patients was surgical treatment, and after having a recurrence at 5 and 4 years, respectively, stereotactic radiosurgery was chosen.

Patient 1

The first patient is a 34-year-old man, who presented nausea and vomiting in 2019 when he got diagnosed.

In August 2019, he underwent surgery for a right cerebellar lesion. After that, he remained without any sign and symptoms after 5 years.

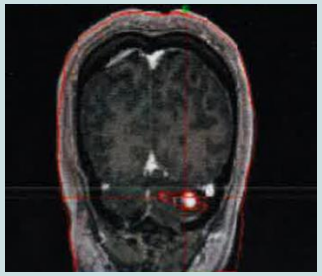


However, a recurrence was found in the left contralateral hemisphere during a follow-up visit.

Patient 2

The second patient, a 32-year-old woman, had a right cerebellar lesion treated surgically in 2020.

Before the procedure, the patient developed hydrocephalus, and a peritoneal ventricle shunt was placed.



She remained without any symptom during the following 4 years, but in 2024 a recurrence was identified, consisting of a three-nodule lesion in the left cerebellar hemisphere.

On procedure day, the female patient's extradural tumor volume was of 2.07 cm3. It received a dose of 20Gy at the margin with a 50% isodose surface and used 5 isocenters: one shot from the 14mm collimator and the other four using the 8mm one.
In the case of the male patient the extradural intraspinal tumor had a target volume of 177.6 cm3. The dose at the margin was 20Gy with a 50% isodose surface and used 1 isocenter, from an 8mm collimator. In both cases the tolerance drop doses in the adjacent critical structures were non-significant.

Follow-up

During the first week of postoperative treatment patients remained asymptomatic. Currently they need to continue with their follow-up consults every 6 months for 2 years, followed by yearly visits for 5 additional years, every time with a new MRI.

Conclusions

This report highlights the infrequent occurrence of the Syndrome of Von Hippel Lindau in two siblings. Furthermore, both patients having a similar progression of treatment and recurrence of the tumors.

References

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2. Hes, F.J., Höppener, J.W., Luijt, R.B.v.d. et al. Von Hippel-Lindau Disease. Hered Cancer Clin Pract 3, 171 (2005). https://doi.org/10.1186/1897-4287-3-4-171