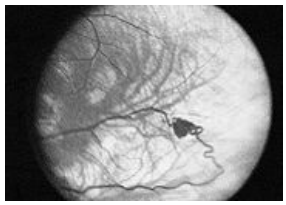
Von Hippel-Lindau syndrome

Von Hippel-Lindau syndrome is an inherited disease of AD in which the VHL gene is mutated (3p 25). The VHL gene product, the VHL protein, binds HIF (hypoxia-inducible factor) factors.



Angiogram with hemangioblastoma

Clinical Picture

🔎 *For more information see Spinal tumors.*

It involves the formation of angiofibromas and hemangioblastomas in the retina and cerebellum. Pheochromocytomas and skin fibromas are also common. About 2/3 of those affected have cysts, they appear in the kidneys, pancreas, and epididymis. There is also a markedly increased risk of developing early renal cell carcinoma.

Diagnostics

Best MRI with Gadolinium contrast. Finding of hemangioblastoma in the cerebellum as a cyst with a hyperdense tumor nodule. USG of kidney, pancreas. Polyglobulins in ectopic erythropoietin production.

Therapy

Therapy is surgical, involving removing tumors and monitoring the patient. Cystic hemangioblastoma in the cerebellum is an indication for urgent surgery. Horseback riding, acute occipital herniation, and death within a few minutes. The cyst must be drained and the tumor nodule extirpated.

Summary video

Links

Related Articles

- Polycystic kidney disease
- Spinal Tumors

Bibliography

- MUDR SOBOTKA, Roman. *Cystická onemocnění ledvin* [lecture for subject Urologie, specialization Všeobecní lékařství, 1.LF UK Univerzita Karlova v Praze]. Praha. 2013.