Teaching NeuroImages: CNS hemangioblastomas in von Hippel-Lindau disease with exon 3 deletion

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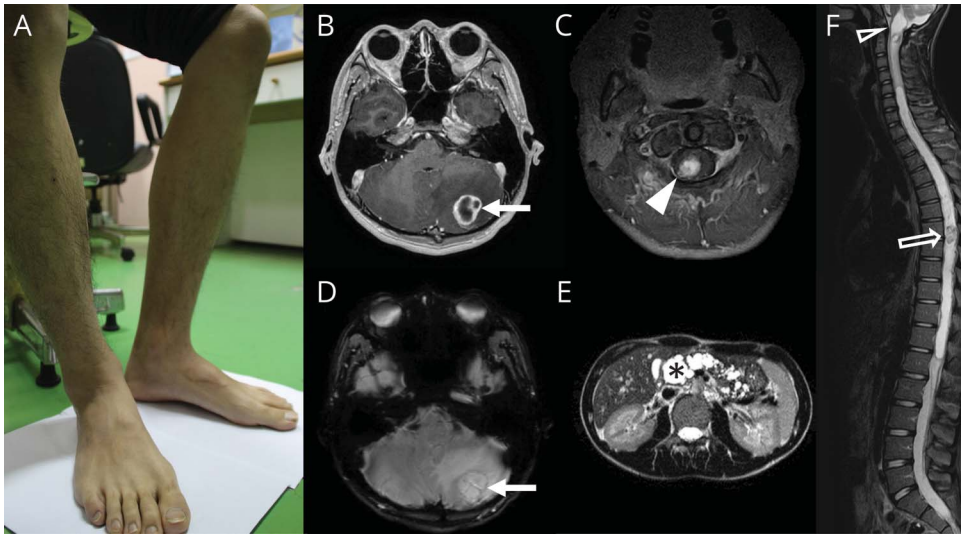
Figure Images of von Hippel-Lindau disease



(A) Calf atrophy. (B, C) Hemangioblastomas at left cerebellum (arrow) and cervicomedullary junction (arrowhead) (enhanced T1-weighted images). (D) Feeding artery (arrow) (gradient echo image). (E) Multiple pancreatic cysts (asterisk) (T2-weighted image). (F) Another hemangioblastoma at T7-8 (open arrow), syringobulbia (open arrowhead), and thoracic syringomyelia (T2-weighted image).

A 14-year-old boy developed progressive bilateral leg weakness for 5 months. Physical examinations showed calf muscle atrophy (figure, A) and hyperreflexia. Spinal lesion of slow progression was suspected.

Spine and brain MRI showed hemangioblastomas at spinal cord and cerebellum, syringobulbia, and thoracic syringomyelia (figure, B–D and F). Abdominal MRI revealed multiple pancreatic cysts (figure, E). The patient also had retinal hemangioblastomas. Genetic analysis showed heterozygous de novo exon 3 deletion of *VHL* gene.

Weakness occurs in 65% of von Hippel-Lindau disease cases.[1] Our patient had truncating mutation, correlating to a higher rate of hemangioblastoma, but a lower risk for pheochromocytoma.[2]

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Author contributions

Drs. Chen and Tsai participated in the neuroimaging interpretations and clinical care of the patient. The manuscript was drafted by Drs. Chao and Tsai and was revised by Dr. Chen.

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