

DIFFICULTIES OF MANAGEMENT OF A VHL FAMILY

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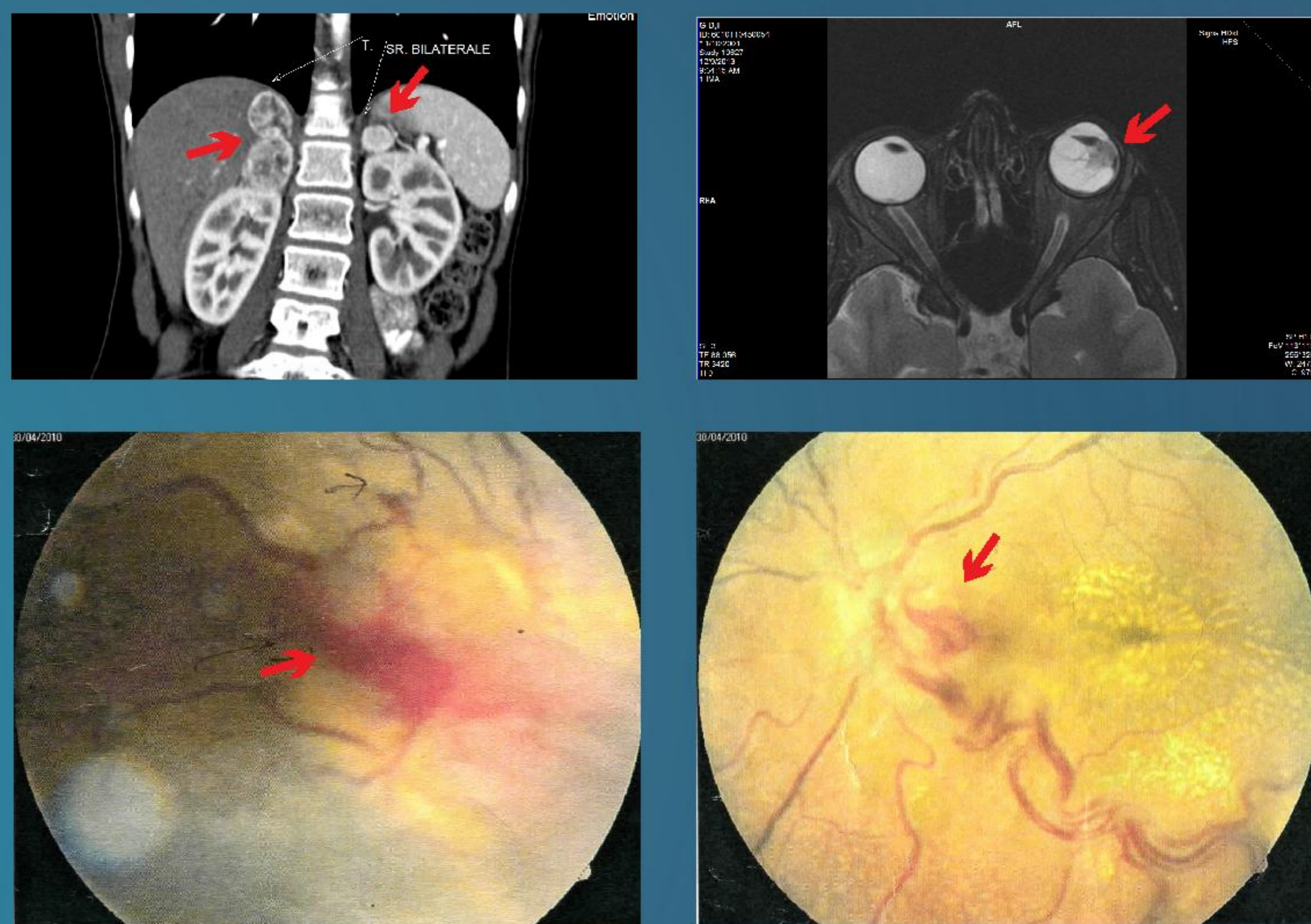
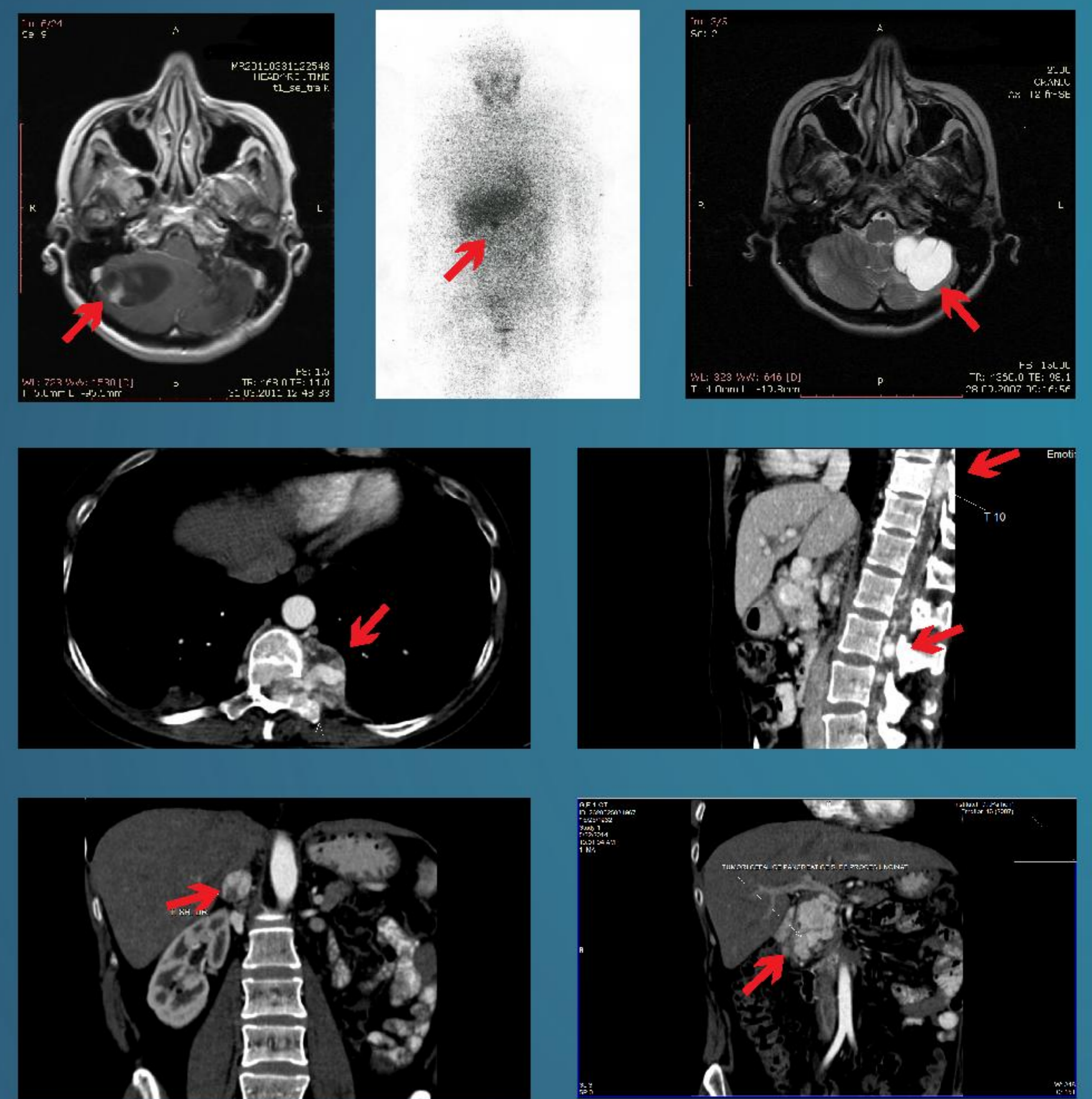
Von Hippel-Lindau (VHL) disease is an inherited, autosomal dominant syndrome manifested by a variety of benign and malignant tumors, with an incidence of about 1 in 36,000 individuals and a mean age at presentation of 26 years. We present a family (mother and daughter) case of possible type 2A VHL disease diagnosed with multiple VHL-associated tumors in the absence of an available genetic testing. They both presented in our clinic in 2013 with biochemical profiles suggestive of pheochromocytoma (PHC). No genetic counseling was given to the mother.

CASE 1

G.F. 54 y.o, known with VHL disease, first diagnosed with bilateral pheochromocytoma (PHC) at the age 13, triple operated- **two bilateral PHC**, in 1976 and 1983, and a **right adrenal tumor (AT) recurrence** - each time a partial adrenal resection was performed - associating **retinal angiomas** with **blindness of the right eye** due to hemoftalmus and multiple **hemangioblastomas of the cerebellum and spinal cord** that required seven neurosurgeries for compressive tumors.

On routine checking we diagnosed **a forth right adrenal recurrence of PHC** of aprox 30/20 mm (normal MN values, **high plasma NMN = 658/837 pg/ml** and normal values of chromogranine A, plasma serotonin, 5-HIAA) in addition to **pancreatic tumors**.

The patient chose to remain under **chronic alpha and beta blockers treatment**.



CASE 2

G.D.I. 15 y.o, prematurely born at 7 months, diagnosed with **retinal angiomas**, complicated cataracts of both eyes and **blindness of the left eye** since she was 9 y.o., was first diagnosed of the age 12 with **bilateral PHC** (2 right AT and a left AT - normal MN values and **high plasma NMN 2842 pg/ml**).

After surgical removal (total left and subtotal right adrenalectomy) one year later, developed a **right adrenal recurrence** for which she underwent total adrenalectomy.

She remains under close surveillance at every three months.

CONCLUSIONS

The management of VHL disease is based upon an **annually multidisciplinary surveillance strategy**, the primary goal being an early diagnosis and avoiding potential tumor-related disability. A regular follow-up with imaging and observation of clinical signs and symptoms starting from the first year of life is needed, never forgetting about the genetic counseling.