

# 24<sup>th</sup> Annual Resident Poster Day

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## A syndrome of pheochromocytomas/paragangliomas (PPGLs) associated with congenital polycythemia and ophthalmic abnormality.

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Pheochromocytomas and paragangliomas (PPGLs) are rare neuroendocrine tumors with the incidence of approximately 0.8 per 100,000 persons. Pheochromocytomas are catecholamine-secreting tumors that arise from the chromaffin cells of the adrenal medulla. Paragangliomas (PGLs) are neuroendocrine tumors that arise from the sympathetic and/or parasympathetic paraganglia and can secrete catecholamines. Pheochromocytomas and paragangliomas are indistinguishable at the cellular level. Approximately 30-35% of PPGLs are reported to be germline mutations, while 35-40% are due to somatic mutations. Most cases present with recurrent PGLs, some with somatostatinomas, some others with ophthalmologic findings, congenital malformation of intracranial veins, or type I Chiari malformations. We report a case of a 44-year-old male with history of congenital blindness, white matter disease likely due to in-utero with cerebral ataxia, polycythemia diagnosed at age 12 previously treated with phlebotomies, and spinal stenosis who presented with new diagnosis of PPGLs. He had episodes of palpitations associated with sweating, anxiety, pulsatile headache and HTN for approximately 8 years. He was admitted in the hospital for one of these episodes at age 39 where he had negative nuclear stress test. He has no family history of neuroendocrine tumors. His tumors were found rather incidentally at age 44 during the evaluations for his back pain with MRI of lumbar spine showing a paraaortic lesion. This case illustrates a rare syndrome of PPGLs, congenital blindness, and polycythemia, likely due to somatic mutations of FIF2a.