

Marques P et al. Significant Benefits of AIP Testing and Clinical Screening in Familial Isolated and Young-onset Pituitary Tumors. J Clin Endocrinol Metab. 2020;105(6):e2247-60. doi: 10.1210/clinem/dgaa040.

CONTEXT: Germline mutations in the aryl hydrocarbon receptor-interacting protein (AIP) gene are responsible for a subset of familial isolated pituitary adenoma (FIPA) cases and sporadic pituitary neuroendocrine tumors (PitNETs).

OBJECTIVE: To compare prospectively diagnosed AIP mutation-positive (AIPmut) PitNET patients with clinically presenting patients and to compare the clinical characteristics of AIPmut and AIPneg PitNET patients.

DESIGN: 12-year prospective, observational study.

PARTICIPANTS & SETTING: We studied probands and family members of FIPA kindreds and sporadic patients with disease onset ≤ 18 years or macroadenomas with onset ≤ 30 years (n = 1477). This was a collaborative study conducted at referral centers for pituitary diseases.

INTERVENTIONS & OUTCOME: AIP testing and clinical screening for pituitary disease. Comparison of characteristics of prospectively diagnosed (n = 22) vs clinically presenting AIPmut PitNET patients (n = 145), and AIPmut (n = 167) vs AIPneg PitNET patients (n = 1310).

RESULTS: Prospectively diagnosed AIPmut PitNET patients had smaller lesions with less suprasellar extension or cavernous sinus invasion and required fewer treatments with fewer operations and no radiotherapy compared with clinically presenting cases; there were fewer cases with active disease and hypopituitarism at last follow-up. When comparing AIPmut and AIPneg cases, AIPmut patients were more often males, younger, more often had GH excess, pituitary apoplexy, suprasellar extension, and more patients required multimodal therapy, including radiotherapy. AIPmut patients (n = 136) with GH excess were taller than AIPneg counterparts (n = 650).

CONCLUSIONS: Prospectively diagnosed AIPmut patients show better outcomes than clinically presenting cases, demonstrating the benefits of genetic and clinical screening. AIP-related pituitary disease has a wide spectrum ranging from aggressively growing lesions to stable or indolent disease course.