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GENETICS

AIP mutations in young patients with sporadic pituitary macroadenoma

Screening for *AIP* mutations in patients <30 years of age with sporadic pituitary macroadenoma could improve clinical management of this population, according to results of a study published in the *European Journal of Endocrinology*.

Mutations in the *AIP* gene, which encodes the aryl hydrocarbon receptor interacting protein, are frequent in patients with familial isolated pituitary adenomas (FIPA) but have also been reported in patients with other pituitary adenoma subtypes. Tumors arising from *AIP* mutations are large (macroadenomas) and occur at a young age.

To evaluate the use of focused genetic screening, Tichomirowa *et al.* studied 163 patients with sporadic pituitary macroadenoma (≥10 mm maximal diameter on MRI) diagnosed when patients were aged <30 years. Other genetic causes or FIPA had been excluded.

Germline *AIP* mutations were found in 11.7% of patients, and DNA sequence changes of uncertain significance or polymorphisms in the *AIP* gene in a further 5.5% of patients. Pathogenic *AIP* mutations were identified in 20.5% of individuals aged <18 years.

Overall, 13.3% of patients with sporadic somatotropinoma, 11.5% of patients with prolactinoma and 6.3% of patients with nonfunctioning pituitary adenoma had *AIP* mutations, whereas no mutations were found in individuals with Cushing disease or thyrotropinoma. Familial screening revealed mutations in the relatives of seven patients with *AIP* mutations, and a microadenoma was diagnosed in two of these relatives.

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Original article Tichomirowa, M. A. *et al.* High prevalence of *AIP* gene mutations following focused screening in young patients with sporadic pituitary macroadenomas. *Eur. J. Endocrinol.* doi:10.1530/EJE-11-0304