Genetic analysis in young patients with sporadic pituitary macroadenomas: besides AIP don't forget MEN1 genetic analysis

Titre	Genetic analysis in young patients with sporadic pituitary macroadenomas: besides AIP don't forget MEN1 genetic analysis
Type de publication	Article de revue
Auteur	Cuny, Thomas [1], Pertuit, Morgane [2], Sahnoun-Fathallah, Mona [3], Daly, Adrian F. [4], Occhi, Gianluca [5], Odou, Marie-Françoise [6], Tabarin, Antoine [7], Nunes, Marie Laure [8], Delemer, Brigitte [9], Rohmer, Vincent [10], Desailloud, Rachel [11], Kerlan, Véronique [12], Chabre, Olivier [13], Sadoul, Jean-Louis [14], Cogne, Muriel [15], Caron, Philippe [16], Cortet-Rudelli, Christine [17], Lienhardt, Anne [18], Raingeard, Isabelle [19], Guedj, Anne-Marie [20], Brue, Thierry [21], Beckers, Albert [22], Weryha, Georges [23], Enjalbert, Alain [24], Barlier, Anne [25]
Editeur	BioScientifica
Type	Article scientifique dans une revue à comité de lecture
Année	2013
Langue	Anglais
Date	2013 Apr
Pagination	533-541
Volume	168
Titre de la revue	European Journal of Endocrinology
ISSN	1479-683X
Mots-clés	Adenoma [26], Adolescent [27], Adult [28], Child [29], Cohort Studies [30], Female [31], Genetic Linkage [32], Humans [33], Intracellular Signaling Peptides and Proteins [34], Male [35], Mutation [36], Pituitary Neoplasms [37], Proto-Oncogene Proteins [38], Young Adult [39]

Résumé en anglais	**CONTEXT:** Germline mutations in the aryl hydrocarbon receptor interacting protein gene (AIP) have been identified in young patients (age ≤30 years old) with sporadic pituitary macroadenomas. Otherwise, there are few data concerning the prevalence of multiple endocrine neoplasia type 1 (MEN1) mutations in such a population. **OBJECTIVE:** We assessed the prevalence of both AIP and MEN1 genetic abnormalities (mutations and large gene deletions) in young patients (age ≤30 years old) diagnosed with sporadic and isolated macroadenoma, without hypercalcemia and/or MEN1-associated lesions. **DESIGN:** The entire coding sequences of AIP and MEN1 were screened for mutations. In cases of negative sequencing screening, multiplex ligation-dependent probe amplification was performed for the detection of large genetic deletions. **PATIENTS AND SETTINGS:** One hundred and seventy-four patients from endocrinology departments of 15 French University Hospital Centers were eligible for this study. **RESULTS:** Twenty-one out of 174 (12%) patients had AIP (n=15, 8.6%) or MEN1 (n=6, 3.4%) mutations. In pediatric patients (age ≤18 years old), AIP/MEN1 mutation frequency reached nearly 22% (n=10/46). AIPmut and MEN1mut were identified in 8/79 (10.1%) and 1/79 (1.2%) somatotropinoma patients respectively; they each accounted for 4/74 (5.4%) prolactinoma (PRL) patients with mutations. Half of those patients (n=3/6) with gigantism displayed mutations in AIP. Interestingly, 4/12 (33%) patients with non-secreting adenomas bore either AIP or MEN1 mutations, whereas none of the eight corticotroph adenomas or the single thyrotropinoma case had mutations. No large gene deletions were observed in sequencing-negative patients. **CONCLUSION:** Mutations in MEN1 can be of significance in young patients with sporadic isolated pituitary macroadenomas, particularly PRL, and together with AIP, we suggest genetic analysis of MEN1 in such a population.
URL de la notice	http://okina.univ-angers.fr/publications/ua10560 [40]
DOI	10.1530/EJE-12-0763 [41]
Titre abrégé	Eur. J. Endocrinol.
Identifiant (ID) PubMed	23321498 [42]

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