

Table 2: Clinical and genetic characteristics of the 15 patients harboring a likely pathogenic or pathogenic variant

N°	Sex	Size of Adenoma	Age at Diagnosis (years)	Family context	Nucleotide nomenclature	Protein nomenclature	Variant classification*	Reference
MEN1 (NM_130799)								
1	M	macro	13		c.249_252 del	p.(Ile85Serfs*85)	PV	Chandrasekharappa et al, Science 1997
2	M	macro	12	Sister: pituitary adenoma	c.402del	p.(Phe134Leufs*51)	PV	Chandrasekharappa et al, Science 1997
3	W	macro	17	0	c.415del	p.(His139Thrfs*15)	PV	Wautot et al, Hum Mutat, 2002
4	W	macro	11	0	c.574 C>T	p.(Gln192*)	PV	Jager et al, Mol Cell Endocrinol, 2006
5	M	macro	15	0	c.788T>G	p.(Leu263Arg)	PPV	Not previously reported
6	M	macro	16	0	c.1546del	p.(Arg516Glyfs*43)	PV	Agarwal et al, Hum Mol Genet, 1997
7	W	macro	15	0	c.1546dup	p.(Arg516Profs*15)	PV	Agarwal et al, Hum Mol Genet, 1997
AIP (NM_003977)								
8	M	macro	22	0	c.55C>T	p.(Gln19*)	PV	Mougel et al, Eur J Endocrinol, 2020
9	W	macro	17	0	c.350del	p.(Gly117Alafs*39)	PV	Tichomirowa et al, Eur J Endocrinol, 2011

10	M	macro	18	Nephew: somatotro ph adenoma; cousin: macroprol actinoma	c.404del	p.(His135Leufs*2 1)	PV	Cazabat et al, Eur J Endocrinol, 2012
11	M	macro	10	0	c.442dup	p.(Leu148Pro*75)	PV	Not previously reported
12	W	macro	27	Sister: macroade noma at 31	c.645+2 T>C	p.(?)	PV	Not previously reported
13	M	macro	21	0	c.805_825 dup	p.(Phe269_His27 5dup)	PV	Leontiou et al, J Clin Endocrinol Metab, 2006
14	M	macro	15	0	c.811C>T	p.(Arg271Trp)	PV	Daly et al, J Clin Endocrinol Metabl, 2007

M, man; W, woman; macro, macroadenoma, defined by a diameter >10 mm; micro, microadenoma, defined by a diameter <10 mm ; *Classification of sequence variants according to the ACMG guidelines; PV: pathogenic variant; PPV: probably pathogenic variant