

Appendix

Table 1. Characterisation of 36 Norwegian patients with APS-I: follow-up after 5–10 years

Family no.	Pat. no.	Sex	Age	Age of debut ¹	Autoantibodies ²	Manifestations ³	Aire mutations
I	1	M	11	3	AADC+	HP(3), C(3), AI, K	c.967–979del13/c.769C>T
I	2	M	14	2	21OH+	HP(5), C(2)	c.967–979del13/c.769C>T
II	3	F	16	0	SCC+, (21OH-), 17OH	A(5), HP(5), C(0), E	c.769C>T/c.1242_1243insA
II	4	F	20	9	21OH, 17OH	A(9)	c.769C>T/c.1242_1243insA
II	5	F	25	14	21OH, 17OH	SCC, A(14), C(14), V, N, G	c.769C>T/c.1242_1243insA
III	6	M	16	4	21OH, TH	A(4), HP(4), C(4), E, M	c.967–979del13/c.967– 979del13
IV	7	M	19	4	21OH, 17OH	SCC, A(11), HP(4), C(9), E	c.967–979del13/c.967– 979del13
IV	8	M	26	9	21OH, GAD	AADC, A(12), HP(9), C, E	c.967–979del13/c.967– 979del13
IV	9	F	30	4	21OH, GAD, TPH	AADC, HP(4), C, AT, V, E	c.967–979del13/c.967– 979del13
V	10	M	22	8	21OH, TPH, TH	AADC, HP(8), C(14), N, E	c.1163_1164insA/c.967– 979del13
VI	11	M	30	0	21OH, AADC	A(10), HP(10), C(0), AI, H, M	c.967– 979del13/c.1244_1245insC
VII	12	F	39	13	21OH, 17OH	SCC, A(14), HP(13), G, AI, C, AT	c.1244_1245insC/ c.1244_1245insC
VIII	13	M	⁵ 35	4	21OH, AADC, TH	SCC, A(4), HP(4), C(26), AI, E, SC	c.967–979del13/c.967– 979del13
IX	14	M	44	20	SCC, GAD, TPH	AADC, A(20), HP(27), C(30)	c.967–979del13/c.967– 979del13
IX	15	F	⁵ 10	9	n.d.	A(9), HP?	n.d.
X	16	F	44	7	21OH	A(22), C(7), V, AI, M, E	Not found
XI	17	F	48	10	21OH, SCC	A(13), HP(10), C(10), G	c.967–979del13/c.967– 979del13
XII	18	F	49	1	21OH, AADC+, TPH, 17OH	SCC, GAD, A(17), HP(15), C(1), G	c.967–979del13/c.967– 979del13
XIII	19	F	54	13	21OH, GAD	A(13), HP(13), C(13), G, AT, AI,	c.769C>T / c.1336T>G

					E, D			
XIV	20	M	58	9	21OH, AADC, GAD+	SCC, A(16), AI, C	HP(9), V,	c.769C>T/c.769C>T
XIV	21	F	⁵ 6	Not known	n.d.	n.d.	n.d.	n.d.
XIV	22	M	⁵ 12	Not known	n.d.	n.d.	n.d.	n.d.
XV	23	M	61	5	21OH	A(17), C(5), K, V, AI	HP(20),	c.967–979del13/c.967–979del13
XVI	24	F	30	1	AADC, GAD, TPH	HP(1), AI, E	C(5), G, N,	c.769C>T/c.769C>T
XVII	25	F	46	9	Not found	HP(9), N, E, C	G(17), AI,	c.22C>T / c.290T>C
XVIII	26	M	36	12	(21OH-), (GAD-)	SCC, A(12)		c.967–979del13/c.967–979del13
XIX	27	M	55	Not known	21OH, SCC	A, HP, AI		c.22C>T/c.402delC
XX	28	F	32	21	21OH, TPH	A, C		c.879+1G>A/c.879+1G>A
XX	29	M	47	43	AADC, GAD, TPH	HP(43), D, V		c.879+1G>A/c.879+1G>A
XXI	30	M	⁵ 49	13	21OH, AADC, TPH	SCC, A, HP, C, AI		c.967–979del13/c.1249dupC
XXII	31	M	42	14	AADC, GAD, TPH	HP(14), K, N, V, AI	C(22), D,	c.769C>T/c.1249dupC
XXIII	32	M	⁵ 39	14	n.d.	A, C		c.967–979del13/c.769C>T
XXIV	33	M	48	Not known	Not found	C, HP		c.967–979del13/c.967–979del13
XXIV	34	F	⁵ 3	3	n.d.	HP		n.d. ⁴
XXIV	35	M	⁵ 17	5	n.d.	A, HP, C, AI, E		n.d. ⁴
XXIV	36	F	⁵ 34	8	n.d.	HP, AT, E		n.d. ⁴

A, Adrenal insufficiency; HP, hypoparathyroidism; C, mucocutaneous candidiasis; G, primary gonadal insufficiency; V, vitiligo; AI, alopecia; AT, autoimmune thyroid disease; M, malabsorption; N, nail pitting; E, dental enamel hypoplasia; K, keratopathy; H, autoimmune hepatitis; SC, oral squamous cell carcinoma; D, diabetes type 1; n.d., not done.

¹The age of debut denotes the age at which the first APS-I main component appeared.

²Autoantigen written in bold with a plus sign signifies that reactivity against a given antigen has appeared, whereas autoantigen written in bold within parentheses with a minus sign denotes that reactivity against a given autoantigen was lost, compared to earlier analyses.

³The year of diagnosis for the manifestation is written in parentheses.

⁴Mother is heterozygot for c.967–979del13.

⁵Deceased (age in years).

Reproduced with permission from Wolff et al, Journal of Clinical Endocrinology & Metabolism 2007 [46]