Supplement A: AADC deficiency: included patients

Pt NR	Core diagnostic tests reported	References	TML	Prolactin	Blood NT	Urine NT	CSF NT	CSF GT
1	CSF, AADC	[1, 2]				x**		
2	CSF, AADC, DNA	[2-4]	x	x	x	x**	x	
3	CSF, DNA	[5]			x			
4	CSF, AADC	[6]			x			
5	CSF, AADC, DNA	[7, 8]		x	x			
6	CSF, AADC, DNA	[8, 9]		x		x	x	
7	CSF, AADC, DNA	[10, 11]		x		x		x
8	CSF, AADC, DNA	[2, 12-16]	x			x**	x	
9	CSF, AADC, DNA	[2, 3, 12-16]	x			x**	x	
10	CSF, AADC, DNA	[2, 3, 12-16]	x			x**	x	
11	CSF, AADC, DNA	[3, 13, 15, 17]				x**	x	
12	CSF, AADC, DNA	[13, 17, 18]	x	x		x**		
13	CSF, AADC, DNA	[19]		x				
14	CSF, AADC, DNA	[20-23]			x	x	x	
15	CSF, AADC, DNA	[20-23]			x	x	x	
16	CSF, AADC, DNA	[11, 24]					x	x
17	CSF, AADC, DNA	[11, 24]					x	x
18	CSF, AADC, DNA	[14, 25]				x	x	
19	CSF, DNA	[26]		x		x**		
20	CSF, DNA	[27, 28]				x**		
21	CSF, DNA	[29]				x**		
22	CSF, AADC, DNA	[30]		x	x	x		
23	CSF, AADC, DNA	[30]		x	x	x		
24	CSF, AADC	[31]			x			
25	CSF, AADC, DNA	[14]				x		
26	CSF, AADC, DNA	[32]				x		
27	CSF, AADC, DNA	[14]				x		
28	CSF, AADC, DNA	[33]				x		
29	CSF, AADC, DNA	[15, 33]				x		
30	CSF, AADC, DNA	[2, 13, 15]			x	x**		
31	CSF, AADC, DNA	[2, 13, 15]	x			x**		
32	CSF, AADC, DNA	[2, 13, 15]	x			x	x	
33	CSF, AADC, DNA	[2, 13, 15]	x		x	x**	x	
34	CSF, AADC, DNA	[2]		x				
35	CSF, DNA	[2]		x		x		
36	CSF, AADC	[2]		x				
37	CSF, DNA	[2]		x				
38	CSF, AADC	[2]		x				
39	CSF, AADC	[2]		x				
40	CSF, AADC	[2]		x				
41	CSF, DNA	[2]		x				
42	CSF, DNA	[2]		x				
43	CSF, DNA	[2]		x				
44	CSF, DNA	[2]		x				
45	CSF	[2]		x		x		

Pt NR	Core diagnostic tests reported	References	TML	Prolactin	Blood NT	Urine NT	CSF NT	CSF GT
46	CSF	[2]		X		x		
47	DNA	[15]				x		
48	DNA	[15]				x		
49	DNA	[15]	x		x	x**		
50	DNA	[15]				x		
51	DNA	[15]				x		
52	DNA	[15]				x		
53	DNA	[15]				x		
54	DNA	[15]				x		
55	DNA	[15]				x		
56	DNA	[34]				x**		
57	CSF, AADC, DNA	[8]				x		
58	CSF, AADC, DNA	[8]				x		
59	CSF, AADC, DNA	[8]				x		
60	CSF, AADC, DNA	[8]				x		
61	DNA	[8]				x		
62	CSF, DNA	[8]				x		
63	CSF, AADC, DNA	[8]				x		
64	CSF, AADC, DNA	[8]				x		
65	CSF, AADC, DNA	[11, 35]					x	x
66	DNA	[36]		x	x*			
67	DNA	[36]		x	x*			
68	DNA	[36]		x	x*			
69	DNA	[36]		x	x*			
70	CSF, DNA	[37]			x*			
71	CSF, DNA	[38]			x*			
72	CSF, DNA	[38]			x*			x
73	CSF, DNA	[38]			x*			x
74	CSF, DNA	[38]			x*			x
75	CSF, DNA	[38]			x*			x
76	CSF, DNA	[38]			x*			x
77	CSF, DNA	[38]			x*			x
78	CSF, DNA	[38]			x*			x
79	CSF, DNA	[38]			x*			x
80	CSF, DNA	[38]			x*			x
81	CSF, AADC, DNA	[11]						x
82	CSF, AADC, DNA	[11]						x
83	DNA	[39]		x				
84	DNA	[39]		x	x*			
85	DNA	[39]			x*			
86	DNA	[39]			x*			
87	DNA	[39]			x*			
88	DNA	[40]			x*	x		
89	DNA	[40]			x*	x		
90	DNA, AADC, CSF	[41]			x*			
91	DNA, AADC, CSF	[41]			x*			
92	DNA, AADC, CSF	[41]			x*			
93	DNA, AADC, CSF	[41]			x*			
94	DNA, AADC, CSF	[41]			x*			
95	DNA, AADC, CSF	[41]			x*			
97	DNA, AADC, CSF	[41]			x*			

Table Legend:

Core diagnostic tests: diagnostic tests that were reported to confirm AADC deficiency. CSF: typical monoamine neurotransmitter profile in cerebrospinal fluid. AADC: low aromatic l-amino acid decarboxylase enzyme activity. DNA: molecular diagnosis confirmed.

TML: Translational Metabolic Laboratory, data included from TML database that was not previously reported in the literature.

Prolactin: serum prolactin reported.

Blood NT: measurements of monoamine neurotransmitter metabolites in blood. *: only 3-OMD measurements available (DBS or plasma)

Urine NT: measurements of monoamine neurotransmitter metabolites in urine. **:measurements of multiple metabolites performed. In the patients without asterix, only dopamine or VLA was reported.

CSF NT: measurement of monoamine neurotransmitter metabolites in cerebrospinal fluid before and during medical treatment

CSF GT: measurement of monoamine neurotransmitter metabolites in cerebrospinal fluid before and after gene therapy.

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