Supplement B: Tyrosine Hydroxylase Deficiency: included patients

Pt NR	Core diagnostic tests	Reference	TML	Prolactin	Urine NT	CSF NT
1	CSF, DNA	[1-4]	x	x	X	x
2	CSF, DNA	[5-7]			X	x
3	CSF, DNA	[5-8]	x		X	x
4	CSF, DNA	[5-8]			X	x
5	CSF, DNA	[5-8]	x		X	x
6	CSF, DNA	[9]		x		
7	CSF, DNA	[9]		x		
8	CSF, DNA	[3, 10, 11]	x	x		x
9	CSF, DNA	[3]			X	
10	CSF, DNA	[12]			X	x
11	CSF, DNA	[13, 14]		x		
12	CSF, DNA	[13, 14]				x
13	CSF, DNA	[13, 14]		x		
14	CSF, DNA	[13-15]		x		x
15	CSF, DNA	[14, 15]		x		x
16	CSF, DNA	[16, 17]				x
17	CSF, DNA	[18]		x	X	x
18	DNA	[19, 20]			X	
19	DNA	[19, 20]			X	
20	CSF, DNA	[21]		x		x
21	CSF, DNA	[22]	x			x
22	CSF, DNA	[14]		x		x
23	CSF, DNA	[14]		x		x
24	CSF, DNA	[23]	x	x	X	
25	DNA	[24]		x		
26	DNA	[24]		x		
27	DNA	[25]			X	

Table Legend:

Core diagnostic tests: diagnostic tests that were reported to confirm TH deficiency. CSF: typical monoamine neurotransmitter profile in cerebrospinal fluid. DNA: molecular diagnosis confirmed.

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

Prolactin: serum prolactin

Urine NT: measurements of multiple monoamine neurotransmitter metabolites in urine

CSF NT: measurement of monoamine neurotransmitter metabolites in cerebrospinal fluid before and during medical treatment (levodopa with peripheral decarboxylase inhibitor).

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