

On-line Table 1: Patient characteristics, imaging studies and infarct detection

Patient	Sex	Mutation Type ^a	Mean Age at Scan (yr)	Length Follow-Up (yr)	No. of Scans	CTA			MRA		3-Vessel Angiogram	Evidence for Infarct on Imaging	Infarct on 1st Available Imaging	Age at 1st Imaging	
						CT	Head	Neck	Head	Neck					
1	F	Nonclassic c.1968 + 1G>A	8.2	0.4	5	5						+	+	8.0	
2	M	Presumed classic	10.8	0	1	1									
3	M	Classic	6.5	1.4	12	4		5	2	1		+	+	5.7	
4	M	Classic	9.4	0.4	4			2	1	1					
5	M	Classic	10.2	0	3			1	1	1		+	+	10.2	
6	M	Presumed classic	11.1	3.4	14	9	1	2	1	1		+		10.7	
7	F	Classic	12.2	0	2			1	1						
8	M	Classic	6.9	10.3	4	2		2				+		10.3	
9	M	Classic	12	2.6	9	1		3	3	2		+	+	10.5	
10	M	Presumed nonclassic	3.9	4.8	8	3		4	1			+	+	4.9	
11	F	Classic	6.7	7.3	5	2		3				+		7.5	
12	F	Classic	2.6	3.8	9	5		4							
13	F	Presumed classic	4.3	0	1	1									
14	M	Classic	0.4	0.5	2	1		1							
15	F	Classic	7.9	1.6	3			3				+	+	6.4	
16	M	Heterozygous c.1822 G>A (G608S)	1.3	0	2			1	1						
17	F	Classic	6.8	2.7	7	1		4	2			+	+	5.8	
18	F	Classic	0.9	1	6			2	2	2		+	+	0.4	
19	F	Classic	6.9	0.5	2			1			1				
20	M	Classic	2.1	0	1	1									
21	M	Classic	9.4	0.1	4	1	1	1				+	+	9.3	
22	M	Classic	7.8	3.8	10	4		3	2		1	+	+	6.1	
23	M	Classic	1.9	2.6	4			3	1			+		1.8	
24	F	Classic	7.2	2.7	7	1		4	1	1		+	+	5.0	
25	F	Heterozygous c.1822 G>A (G608S)	0.4	0.4	1			1							
Column Totals						126	42	2	1	51	19	9	2	15	11

Note:—+ indicates present.

^a Classic indicates a heterozygous c.1824 C>T (G608G) mutation.

On-line Table 2: Neurovascular imaging

	Present (No.) (% Visualized)	Absent (No.)	N/V (No.)
Vessel characteristics			
Cervical CCA stenosis	1 (10%)	9	15
Cervical ICA stenosis	3 (30%)	7	15
CCA, cervical ICA, ECA calcification	1 (50%)	2	12
Proximal cervical VA stenosis	1 (11%)	8	16
Cervical VA calcification	1 (10%)	9	16
Distal V2/V3 segment VA stenosis/occlusion	9 (82%)	2	14
Precavernous ICA stenosis/occlusion	11 (85%)	2	12
Cavernous ICA stenosis/occlusion	12 (92%)	1	12
Supraclinoid ICA stenosis/occlusion	7 (54%)	6	12
ACA stenosis/occlusion	8 (62%)	5	12
MCA stenosis/occlusion	6 (46%)	7	12
PCA stenosis/occlusion	2 (14%)	12	11
BA stenosis/occlusion ^a	0 (0%)		
V4 segment VA stenosis/occlusion ^a	0 (0%)		
Vessel prominence due to collateralization			
Cortical collateral venous flow	14 (67%)	7	4
Suprasellar cistern collaterals	12 (63%)	7	6
Subfrontal collaterals	11 (58%)	8	6
Perisplenic collaterals	2 (15%)	11	12
Enlarged MMA	5 (45%)	6	14
Enlarged IMA	6 (55%)	5	14
Enlarged deep cervical arteries	7 (78%)	2	16
Enlarged occipital arteries	7 (70%)	3	15
Enlarged ascending cervical arteries	4 (40%)	6	15
Enlarged ASA	8 (80%)	2	15
Enlarged posterior spinal artery	3 (30%)	7	15

Note:—N/V indicates feature not well-visualized; BA, basilar artery; ECA, external carotid artery; IMA, internal maxillary artery; MMA, middle meningeal artery.

^a Not identified, but no overall number noted