

eTable 2: Correlation between alternate allele frequency of pathogenic somatic mutations in mTOR pathway genes and histological findings.

Patient Gene, variant	Sample ID	Location	Histology	Alternate allele fraction	(alt/total read count)
Patient #1 <i>MTOR</i> : c.4448G>A p.Cys1483Tyr	Surgery #1, R orbito-frontal and insular resection				
	Fresh brain	Right fronto-temporal	FCD type IIb	8.5%	8221/106063
	Blood	Not available			
	Saliva	-	-	Not detected	-
	FFPE-2a	Right lateral orbitofrontal	FCD type IIa	3.0%	2459/82334
	FFPE-2b	Right insular cortex	FCD type IIa	11.6%	13671/117713
	FFPE-2c	*lesion	FCD type IIb	3.9%	7978/181560
	Surgery #2, R fronto-temporal resection				
	FFPE- a1	R frontotemporal lobe	FCD type IIa	11.6%	23130/200084
	FFPE-c1	R frontal lobe (cavitron)	FCD type IIa	3.1%	3953/125724
	FFPE-d1	R temporal lobe (cavitron)	FCD type IIa	6.8%	11681/171025
	FFPE- f1	R temporal lobe	FCD type IIa	10.3%	9345/90735
	FFPE- 1h	R hippocampus	FCD type IIa	3.3%	3323/99669
	Surgery # 3- Right fronto-temporal resection with callosotomy				
	FFPE-B1	Right frontal lobe	FCD type IIa	4.0%	1784/44938
	FFPE-B2	Right frontal lobe	FCD type IIa	3.9%	2919/75889
	FFPE-B3	Right frontal lobe	FCD type IIa	5.0%	2818/56732
	FFPE-B4	Right frontal lobe	FCD type IIa	3.5%	1557/45045
FFPE-B5	Right frontal lobe	FCD type IIa	3.7%	2282/62247	
Patient # 2 <i>MTOR</i> : c.7499T>A, p.Ile2500Asn	Surgery 1#, Right medial occipital epileptogenic zone				
	Fresh brain	Right medial occipital lobe	FCD type IIa	4.4%	3753/85670
	FFPE-A	Right occipital epileptogenic zone	FCD type IIa	5.4%	4138/77085
	FFPE-D	Right medial occipital epileptogenic zone	FCD type IIa	5.4%	5609/104700
	FFPE-E	Right occipital pole	FCD type IIa	1.3%	1663/129077
	Surgery # 2- Functional right hemispherectomy				
	Fresh brain	Right anterior temporal pole	FCD type IIa	7.6%	7159/94707
	FFPE-A	Right anterior temporal lobe	FCD type IIa	6.2%	3755/61005
	FFPE-B	Parahippocampal gyrus	FCD type IIa	1.2%	837/71411
	FFPE-D	Inferior parietal lobe	FCD type IIa	1.2%	642/51680
Patient # 3 <i>MTOR</i> : c.4373_4375dupATG, p.Asp1458dup					
	Fresh brain (694)	Posterior cingulate	FCD type IIa	1.3%	1575/117532
	Fresh brain	Posterior cingulate	FCD type IIa	3.1%	80/2552
	blood	-	-	Not detected	-
Patient # 4	Surgery #1, R parieto-occipital resection				

<p><i>MTOR: c.4447T>C</i> <i>p.Cys1483Arg</i></p>	Fresh brain	Right parieto-occipital lobe	FCD type IIb	1.12%	1126/100974	
	Blood	-		Not detected	-	
	Saliva	-		Not detected	-	
	FFPE- 1a	Right parieto-occipital lobe	Normal	Not detected	-	
	FFPE- 1b	Right parieto-occipital lobe	Normal	1.2%	778/65374	
	FFPE- 1c	Right parieto-occipital lobe	Normal	0.6%	158/25706	
	FFPE- 1d	Right parieto-occipital lobe (cavitron)	Ectopic neurons	1.1%,	3100/283224	
	Surgery #2, R anterolateral frontal lobe resection					
	FFPE- 1e	Right anterolateral frontal lobe	FCD type IIa	8.8%	7836/89062	
	FFPE- 1f	Right anterolateral frontal lobe (cavitron)	FCD type IIb	5.9%	6600/111513	
	Surgery #3, R orbitofrontal lobe resection					
	FFPE- 1h	Right orbitofrontal gyrus	FCD type IIb	1.1%	1126/100974	
	FFPE- 1i	Right superior frontal gyrus	Few dysmorphic neurons	1.1%	126/11550	
	FFPE- 1j	Right inferior frontal gyrus	Few dysmorphic neurons	0.8%	1045/126189	
FFPE- 1k	Right orbitofrontal gyrus	FCD type IIb	3.6%	720/20280		
FFPE- 1l	Right frontal lobe (cavitron)	FCD type IIb	3.6%	3480/98150		
<p>Patient # 5 <i>MTOR: c.6644C>A</i> <i>p.S2215Y</i></p>	Right frontal lobe resection					
	Fresh brain	Right Frontal lobe (B), lesion	FCD type IIb	1.9%	708/37493	
	Blood	-	-	Not detected	-	
	Saliva	Not available				
	FFPE-A	Right frontal lobe, anterior border of dysplasia	Gliosis	Not detected	-	
	FFPE-C	Right frotal lobe- posterior border of dysplasia	Gliosis	Not detected	-	
FFPE-D	Right frontal lobe, cavitron	Few dysmorphic neurons	Not detected	-		
<p>Patient # 6 <i>MTOR: c.6644C>T,</i> <i>p.Ser2215Phe</i></p>						
	Fresh brain	Left parahippocampus	FCD type IIa	0.9%	15/1579	
	Blood	Not available				
	Saliva	Not available				
	FFPE- A1	Left temporal lobe T2-T3 cortex convolutions	FCD type IIa	5.4%	2119/38922	
	FFPE- A2	Left temporal lobe T2-T3 cortex convolutions	FCD type IIa	2.1%	957/45298	
	FFPE- C	Anterior hippocampus	Normal	Not detected	-	
FFPE- D	Posterior hippocampus	Normal	Not detected	-		
<p>Patient # 7 <i>MTOR: c.5930C>A</i> <i>p.Thr1977Lys</i></p>	Surgery #1, Left frontal resection					
	Fresh brain	Left frontal FCD	FCD type IIb	0.8%	275/2062226	
	Blood	-	-	Not detected	-	
	Saliva	Not available				
	FFPE- 3a	Lateral border of suspected FCD	Few dysmorphic neurons	Not detected	-	
	FFPE- 3b	Posterior border of suspected FCD	Normal	Not detected	-	
	FFPE- 3c	Base of sulcus with suspected FCD	FCD type IIb	3.4%	1514/45138	
FFPE- 3d	Base of sulcus with suspected FCD	FCD type IIb	3.1%	1176/38063		

	FFPE- 3e	Cavitron aspirate	FCD type IIb	2.5%	1033/41305
Patient # 8 MTOR: c.6644C>T p.Ser2215Phe	Right orbitofrontal resection				
	Fresh brain	Right orbitofrontal lesion	FCD type IIa	1.4%	400/27619
	Blood	-	-	Not detected	-
	FFPE	Cavitron aspirate	FCD type IIa	Not detected	-
Patient # 9 MTOR: c.5930C>A, p.Thr1977Lys	Left inferior post-central gyrus and anterior supramarginal gyrus resection				
	Fresh brain	M. 3/4 OF THE WAY TO LESION ELECTRODE 2 AND 3 SLIGHTLY SUPERIOR	FCD type IIb	0.7%	318/43056
	Blood	-	-	Not detected	-
	Saliva	Not available			
	FFPE-B	Towards the lesion	Normal	Not detected	-
	FFPE-C	Anterior to lesion (between electrodes 2 and 3)	Few dysmorphic neurons	Not detected	-
	FFPE-F	Superior aspect of lesion (electrode 5)	Few dysmorphic neurons	Not detected	-
	FFPE-G	Midway to lesion electrode 2	FCD type IIa	Not detected	-
	FFPE-H	Midway to lesion electrode 3	Few dysmorphic neurons	Not detected	-
	FFPE-J	Superficial and posterior to lesion, electrode 4	Few dysmorphic neurons	Not detected	-
	FFPE-M	¼ of the way to lesion, electrode 2 and 3, slightly superior (<i>*note variant detected in mirror specimen from fresh-frozen tissue</i>)	FCD type IIb	Not detected	-
	FFPE-O	depth of lesion, close to insula (depth of electrode 2 and 3)	Few dysmorphic neurons	Not detected	-
Patient # 10 TSC2: c.2356-1G>A	Parieto-occipital lobectomy				
	Fresh brain	Left parietal tuber	FCD type IIb	9.4%	306/3269
	Blood	-		6.3%	286/4527
	Saliva	-		3.6%	227/6258
	FFPE- A4	Posterior left parietal tuber	FCD type IIb	6.4%	325/5057
	FFPE- A7	Anterior left occipital tuber	FCD type IIb	7.0%	299/4297
FFPE- A11	Anterior left occipital tuber	FCD type IIb	8.0%	124/1556	
Patient # 11 AKT3: c.49G>A, p.Glu17Lys	Left fronto-parietal lobectomy				
	Fresh brain	Left temporal lobe	FCD type IIa	4.9%	74/1508
	Blood	Not available			
	Saliva	-	-	Not detected	
	FFPE- D3, #1006	Insular cortex	FCD type IIa- less marked anomalies	1.3%	12/910
	FFPE- A3	Temporal lobe	FCD type IIa	11.0%	285/2541
FFPE- C5	Amygdala and hippocampus	Hippocampal sclerosis and rare dysmorphic neurons	Not detected		
Patient # 13	Right frontal lobe resection				

PIK3CA: c.1624G>A, p.Glu542Lys	Fresh brain	Right frontal lobe polymicrogyria	FCD type IIa	12%	167/1386
	Blood	Not available			
	Saliva	Not available			
	FFPE-A1	Right frontal lobe polymicrogyria	FCD type IIa	22.7%	16569/73038
	FFPE-A2	Right frontal lobe polymicrogyria	FCD type IIa	20.4%	12652/62001
	FFPE-A3	Right frontal lobe polymicrogyria	FCD type IIa	17.1%	10960/64063
	FFPE-A4	Right frontal lobe polymicrogyria	FCD type IIa	5.1%	4427/87529