

Table 1 Clinical and laboratory features of the 8 patients with MUSD

Patients	1	2	3	4	5	6	7	8
Sex	F	F	M	F	M	M	F	M
Age of Onset	7d	1m	4m	10d	2d	3d	17d	5d
Clinical manifestation								
Vomiting	+	-	-	+	-	-	-	-
Poor response	+	-	-	-	+	+	+	+
Coma	+	-	-	+	-	-	-	-
Convulsion	+	+	-	-	-	+	+	+
Feed difficulties	-	+	-	+	+	+	+	+
Hypermyotonia	-	+	-	-	-	+	+	-
Maple syrup odor	-	+	-	-	-	+	-	-
Mental retardation	-	-	+	-	-	-	-	-
Motor development delay	-	-	+	-	-	-	-	-
Seizure	-	-	-	-	+	-	-	-
Cranial CT/MRI	/	Extensive Low-densi ty changes in the brain	Bilateral Frontotempo ral parietal white mater density symmetry decreased	/	/	/	Poor myelination in white matter, diffuse long T1 and long T2 Edema signal changes in subcortical white matter and frontotempor al white matter	/
Blood tandem mass spectrometry								
Leucine/Isoleucin ($\mu\text{mol/L}$)	1904 (<295)	935 (<200)	1122.88 (<375)	679 (<230)	3089 (<375)	3209 (<200)	1754 (<200)	2748 (<300)
Valine	354 (<270)	295 (<210)	325.05 (<325)	420 (<230)	578 (<325)	521 (<280)	302.92 (<280)	520 (<300)
Leucine/Phenylalanine	34.2 (<7)	19.4 (<8)	29.92(<8)	12.6 (<7)	38.2(<8)	39.1(<8)	22(<8)	29.5 (<8)
Clinical phenotype	Classic	ITD	ITD	ITD	Classic	Classic	ITD	Classic
outcome	Died at 16d	Died at 2m	Mental retardation	Died at 20d	Died at 1m	Died at 10d	Mental retardation	Died at 1m

Notes: F=Female, M=Male, d=day, m=month, ITD =Intermediate, ITM =Intermittent

Table 2:Detection and analysis of gene mutation in 8 MSUD families

Pedigree	Gene	Proband genotype		Paternal genotype	Maternal genotype	Pathogenicity assessment based on ACMG guideline
		Genotype 1	Genotype 2			
1	<i>BCKDHA</i>	/	/	p.Gly281Arg [#]	p.Gly281Arg	PM3+PP3
2	<i>BCKDHA</i>	/	/	p.Ala216Val	p.Ala216Val	
3	<i>BCKDHB</i>	p.Ser171Pro	p.Arg183Trp	p.Arg183Trp	p.Ser171Pro [#]	PM3+PP3
4	<i>BCKDHB</i>	/	/	p.Phe175Leu	p.Lys222Thr [#]	PM3+PM5+PP3
5	<i>BCKDHB</i>	/	/	p.Gly95Ala [#]	p.Arg285Ter	PM3+PP3
6	<i>BCKDHB</i>	/	/	p.Arg111Ter	p.S184Pfs*46	
7	<i>BCKDHB</i>	p.Arg170Cys	p.I160Ffs*25	p.Arg170Cys	p.I160Ffs*25	
8	<i>DBT</i>	/	/	p.Arg431Ter	p.Arg431Ter	