

**Supplementary Table 1:** Clinical features<sup>a</sup> of patients with *HGSNAT* mutation in comparison to features seen in MPS III (including all types A to D)<sup>a</sup> in general.

	HPO <sup>a</sup> ID	Phenotypes of MPS III (types A-D)	Female (III:01)	Male (III:04)
Age at last assessment [years of age]			[32]	[20]
Mode of inheritance		AR	AR	AR
<b>Hair</b>				
Hirsutism	0001007	+	+	-
<b>Bowel and bladder control</b>				
Bowel incontinence	0002607	[+]	+	-
Urinary incontinence [age of onset; years]	0000020	[+]	+	+ [10]
<b>Neurologic signs and symptoms</b>				
Global developmental delay	0001263	+	+	+
Intellectual disability, progressive [age of onset; years]	0006887	+ [1-3]	+	+
Intellectual disability (IQ 70-75 or below)	0001249	+	+ [9 y]	-
Intellectual disability, profound (IQ 19 or below)	0002187	+	+ [13 y]	+
Memory impairment	0002354		+	+
Loss of speech [age of onset; years]	0002371	+	+ [18]	+ [21]
Brain atrophy [MRI; years of age]	0012444	+	+ [30]	+ [24]
Facial diplegia [age of onset; years]	0001349		+ [9]	-
Unsteady gait [age of onset; years]	0002317	+	+ [28]	-
Babinski sign [years of age]	0003487	+	+ [30]	-
Seizures [age of onset; years]	0001250	+	+ [30]	-
Epileptiform EEG discharges [years of age]	0011182	+	-	+ [11]
Hearing impairment	0000365	+	-	+
Abnormal auditory evoked potentials	0006958		n.d.	+
Pain insensitivity [years of age]	0007021		+ [27]	-
Poor motor coordination	0002275	+	+	+
Abnormality of somatosensory evoked potentials	0007377		+	n.d.
Abnormality of visually evoked potentials	0000649	[+]	n.d.	+
Abnormal mitochondria in muscle tissue [years of age]	0008316		n.d.	+ [11]
<b>Behavioral and psychiatric symptoms</b>				
Apathy	0000741	+	n.d.	+
Emotional lability	0000712		+	+
Restlessness [age of onset; years]	0000711	+ [5-10]	+ [4]	+
Disinhibition	0000734		+	+
Aggressive behavior	0000718	+	+	+
Temper tantrums	-	+	+	+
Loss of independence for self-care [age of onset; years]	-		+ [32]	+
Inability to recognize the emotional significance of objects (psychic blindness)	-		+	+
Inappropriate sexual behavior [years of age]	0008768		+ [13] <sup>b</sup>	-
Placidity/tameness	-	[+]	+	+
Polyphagia	0002591		+	+
Hyperorality and the ingestion of inappropriate objects	-		+	+
Excessive tendency to attend and react to visual stimulus (hypermetamorphosis)	-		+	+
Behavioral abnormality [age of onset; years]	0000708	+ [1-6]	+ [<9]	+
Stereotypic behavior	0000733	+	+	+
Impaired social interactions	0000735	+	+	+
Sleep disturbance [age of onset; years]	0002360	+	+ [<9]	+ [11]

<sup>a</sup>All symptoms were listed according to the nomenclature/systematics of the OMIM "Clinical synopsis" and the Human Phenotype Ontology <sup>1</sup>. Phenotype of MPS III has been listed according to <sup>2-6</sup>; <sup>b</sup> Not observed from the age of 28 years onward. Abbreviations: **AR**, autosomal recessive; **EEG**, electroencephalogram; **HPO**, Human Phenotype Ontology; **MPS**, mucopolysaccharidosis; **MRI**, magnetic resonance imaging; **n.d.**, not determined

## SUPPLEMENTARY REFERENCES

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