

Supplementary table Symptom manifestations in IHPRF1 patients

Family	Current report	Gal et al.				Al Sayed et al.						Koroglu et al.				Bramswig et al.										Takenouchi et al.				Campbell et al.		Angus et al.			Bourque et al.												
	Patient	Family 1		Family 1		Family 1		Family 2		Family 2		Family 2		Family 3		Family 4		Family 5		Family 6		Family 7		Family 8		Family 9		Family 10		Family 11		Family 12		Family 1		Family 2	Family 1	Family 1									
	Sex	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male								
Hypotonia*	Severe	Severe	Severe	Severe	Severe	Severe	Mild (improved with age)	Mild (improved with age)	Mild (improved with age)	Severe	Severe	Mild	Mild	Mild	Mild	Mild	Moderate	Severe	Severe	Severe	Severe	Severe	Moderate	+	+	Severe	Severe	Mild	Mild	Mild	Mild	Mild	Mild	Mild	Mild	Mild	Mild	Severe									
Vision	Strabismus	Strabismus	Strabismus, Nystagmus	Strabismus, Ophthalmoplegia	Strabismus	Anisometropia	Normal	Normal	Normal	Strabismus	Strabismus, Nystagmus, Optic atrophy	Strabismus, Nystagmus, Optic atrophy	Strabismus	Strabismus	Strabismus	Strabismus	Strabismus	Strabismus	Normal	Normal	Normal	Strabismus	Strabismus	Strabismus	Strabismus	Strabismus	Strabismus	Esotropia	Esotropia, Myopia	Strabismus, Hypermetropia	Strabismus, Hypermetropia	Normal	Strabismus, Hypermetropia	Strabismus, Hypermetropia	Strabismus, Nystagmus												
Eye Contact	Poor	NA	Absent	Present	NA	NA	NA	Delayed	NA	NA	Absent	Absent	NA	NA	NA	NA	NA	Present	Poor	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA									
Speech Development	+	NA	NA	Delayed	NA	NA	NA	Delayed	NA	NA	NA	NA	NA	NA	NA	NA	Mildly delayed	Delayed	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA									
Scapula	+	NA	NA	+	NA	NA	NA	+	NA	NA	NA	NA	NA	NA	NA	NA	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+								
Constipation	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+								
Muscle Wasting	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+								
Seizure	-	-	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+									
Sleep Disturbance	+	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+								
Cardiovascular Malformation	ASD,TR	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	ASD	NA	NA	NA	NA	NA	NA	NA	NA									
Recurrent Infection	+	NA	NA	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+								
Breathing	Abnormal	Normal	Abnormal	Abnormal	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA								
Neuroimaging	Frontal lobe atrophy	Normal	High intensity foci on dorsal brainstem	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Cerebellar atrophy	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal								
Blood Tests	Abnormal	Normal	Normal	NA	Normal	Normal	Normal	Normal	Normal	Normal	NA	NA	NA	Normal	NA	NA	NA	Intermittently increased TSH	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA									
EEG	Abnormal (Hypsarrhythmia)	NA	NA	NA	NA	Normal	Normal	Intermittent activity with slow background	Intermittent activity with slow background	Normal	Normal	Diffusely slow	Epileptic	NA	NA	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal								
Cognitive Delay*	Severe	Severe	Severe	Severe	Severe	Severe	Severe	Moderate	Moderate	Moderate	Severe	Severe	Severe	Moderate	Moderate	Severe	Severe	Severe	Severe	Severe	Severe	Severe	Severe	Severe	Severe	Severe	Severe	Severe	Moderate	Moderate	Moderate	Moderate	Moderate	Moderate	Moderate	Moderate	Moderate										
Dysmorphisms	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+									
Early Death	-	20 Months	-	5 Years (Due to infection)	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-									
Mutation	c.2563C>T p.Arg855Ter		c.3390G>A IVS29-1G>A			c.1488delT p.Tyr497Thrfs*21		c.3860G>T p.Trp1287Leu		c.1924C>T p.Gln642Ter		mat: c.302C>T p.Arg1008Ter del: c.2629delC p.Gln877Asnfs*16		c.3056dupT p.Leu1019fs*30		mat: c.4281C>A p.Phe1427Leu del: c.4103-2T>C		c.3556C>T p.Gln1186Ter		c.2495dupA p.Glu813Glyfs*23		c.2889+1_dellAAGT p.Val956_Leu963del (24 bp deletion in cDNA)		c.4190C>T p.Arg1384Ter		c.321G>A p.Top107Ter		c.2758delA p.Ile920Leufs*7		4X_6M	c.2671delG p.Val815Serfs*2		5Y_6M		c.537delG p.Trp179Ter		c.1267-2A>G		c.2022_2023delAT p.Cys675Leufs*23		mat: c.110G>A p.Trp377Ter del: c.2723C>T p.Pro908Leu		mat: c.3281G>A P.Arg1094Gln del: c.2496_2497insTCATA p.Phe833Hisfs*40		mat: c.3823C>T p.Arg1275Ter del: c.2496_2497insTCATA p.Phe833Hisfs*40		c.3823C>T p.Arg1275Ter

Note: The extent of the severity of the symptoms marked by \* are results of our own interpretation of the information provided by the respective authors.