

Table S1. List of the 101 genes analyzed by the STP v3 panel.

Gene #	gene symbol
1	A4GALT
2	ABCA1
3	ABHD5
4	ACER1
5	AGA
6	AK1
7	ANK1
8	APOE
9	AP3B1
10	ARSA
11	ARSB
12	ASAH1
13	ASAH2
14	BIN1
15	CHIT1
16	CLCN7
17	CLN3
18	CLN5
19	CLN6
20	CLN8
21	CTNS
22	CTSA
23	CTSK
24	EPB41
25	EPB42
26	FUCA1
27	G6PD
28	GAA
29	GALC
30	GALNS
31	GATA1
32	GBA
33	GBA2
34	GBA3
35	GLA
36	GLB1
37	GM2A
38	GNPTAB
39	GNPTG
40	GNS
41	GPI
42	GRN
43	GUSB
44	HEXA
45	HEXB
46	HGSNAT
47	HYAL1
48	IDS

Gene #	gene symbol
49	<i>IDUA</i>
50	<i>ITCH</i>
51	<i>KIT</i>
52	<i>LAMP1</i>
53	<i>LAMP2</i>
54	<i>LAMP3</i>
55	<i>LIPA</i>
56	<i>LRP2</i>
57	<i>LRRK2</i>
58	<i>LYST</i>
59	<i>MAN2B1</i>
60	<i>MANBA</i>
61	<i>MCOLN1</i>
62	<i>MRC1</i>
63	<i>MTX1</i>
64	<i>NAGA</i>
65	<i>NAGLU</i>
66	<i>NEU1</i>
67	<i>NPC1</i>
68	<i>NPC2</i>
69	<i>NPR2</i>
70	<i>NT5C3A</i>
71	<i>PARK7 (DJ1)</i>
72	<i>PIEZO1</i>
73	<i>PINK1</i>
74	<i>PKLR</i>
75	<i>PPT1</i>
76	<i>PRF1</i>
77	<i>PRKN (PARK2)</i>
78	<i>PSAP</i>
79	<i>RAB27A</i>
80	<i>SCARB2</i>
81	<i>SGSH</i>
82	<i>SLC17A5</i>
83	<i>SLC4A1</i>
84	<i>SMPD1</i>
85	<i>SNCA</i>
86	<i>SNX10</i>
87	<i>SORT1</i>
88	<i>SPTA1</i>
89	<i>SPTB</i>
90	<i>STX11</i>
91	<i>STXBP2</i>
92	<i>SUMF1</i>
93	<i>TCIRG1</i>
94	<i>TFEB</i>
95	<i>TMEM175</i>
96	<i>TNFSF11</i>

[illegible]

STP Panel v 3

Lysosomal storage disorders	49
Erythrocyte membrane disorders	6
Hemolytic anemia	6
Other Hematological Disorders	8
Other disorders	8
Involved in lysosomal function	17
Biomarkers	1
Parkinson disease	6
Total	101

Table S2. Genotype-phenotype summary

Patient	Homozygous variants		Clinical features	MRI findings	Arylsulfatase A activity*
	DNA	Protein			
PET170	c.854+1dup	p.(Pro286Thrfs*2)	Psychomotor regression at 18 months old Hypotonia Peripheral neuropathy Ataxia	T2 and FLAIR hypersignals in white matter Hypoplasia of corpus callosum Ventricles and cerebellar abnormalities	15.21
PET173	c.640G>A	p.(Ala214Thr)	Psychomotor regression at 30 months old	T2 and FLAIR hypersignals in periventricular white matter and semioval center T2 hypersignals affecting corpus callosum Normal ventricles and cerebellum	26.25
PET174	c.640G>A	p.(Ala214Thr)	Psychomotor regression at 36 months old	FLAIR hypersignals in periventricular white matter	15.42

* Normal reference values: 35.16-177.12 (units: nmol/mL/4 hours)

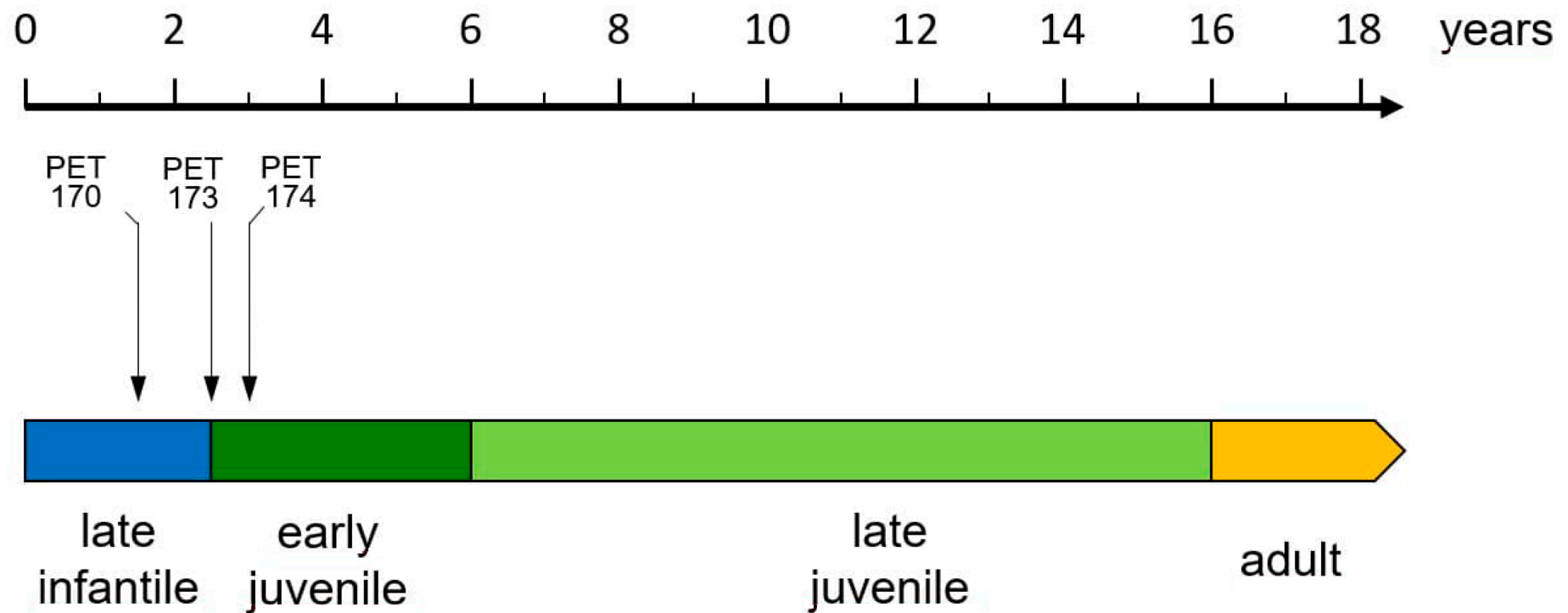


Figure S1. Timeline of onset of psychomotor regression in the three patients described in this work, PET170 (homozygous for c.854+1dup, with earliest onset) from family 1 and PET173 and PET174 (both homozygotes for c.640G>A) from family 2.