Class	Individual		Short stature (<-2.5 SD)	Gestational age > 41 weeks	Neonatal problems	Hypotonia	Feeding difficulties	GERD	Breathing problems	Hypersomnolence	Temperature instability	Excessive hiccups in utero	Neurological abnormality	ID/DD	Stereotypic movements	Hypotonia	Epilepsy	Movement disorder	Delayed myelination	Other brain abnormalities	Skeletal abnormality	Scoliosis	Hip dysplasia	Hyperlaxity	Gastrointestinal abnormality	Swallow problems/drooling	Constipation	Respiratory abnormality	Heart abnormality	Urogenital abnormality	Opthalmologic abnormality	Endocrine abnormality	Vitamin D deficiency	Other	Skin abnormality
A1	2	p.(Gln79*)	-	+	+	+	+	-	+	+	-	U	+	+	-	+	+	-	+	-	+	+	-	+	+	-	+	-	-	+	-	-	-	-	-
	7	p.(Glu9*)	-	-	+	+	+	-	+	+	+	U	+	+	-	+	-	-	-	-	-	-	-	-	+	-	+	+	-	+	-	-	-	-	-
	11	p.(Gly115Profs*87)	-	+	+	+	+	-	+	+	U	+	+	+	-	+	-	-	-	+	-	-	-	+	+	+	+	-	-	+	+	+	U	-
	13	p.(Gly53Alafs*147)	-	-	+	+	+	-	-	+	-	-	+	+	-	+	-	-	-	-	-	-	-	-	+	+	+	-	-	-	+	U	U	U	-
	16	p.(Ile118Hisfs*83)	-	+	+	+	-	-	-	-	-	U	+	+	U	+	-	-	+	-	-	-	-	-	-	-	-	-	-	-	-	U	U	U	-
	18	p.(Leu114Trpfs*111)	U	+	+	+	+	+	-	-	-	U	+	+	U	+	+	-	-	-	-	-	-	U	-	-	-	-	-	+	-	-	-	-	U
	22	p.(Ser43Alafs*34)	+	-	+	+	+	+	+	-	-	U	+	+	U	+	+	U	-	+	-	-	-	U	+	U	+	+	+	-	+	U	U	U	U
	23	p.(Gln128*)	-	+	+	+	+	+	+	-	-	U	+	+	-	+	+	-	-	-	+	+	U	+	+	+	+	+	-	+	-	+	+	+	+
	24	p.(Leu54Cysfs*24)	+	-	+	+	+	+	+	+	+	U	+	+	-	-	+	-	-	-	+	+	+	-	+	+	+	-	+	-	+	-	-	-	U
	26	p.(Leu160*)	-	-	+	+	-	-	-	-	-	U	+	+	-	+	-	+	-	+	+	-	-	+	-	-	-	-	-	-	-	-	U	-	-
	29	p.(Gly47Argfs*155)	-	-	+	+	-	-	-	+	-	-	+	+	-	+	-	-	-	-	-	-	-	+	+	+	-	-	-	-	U	U	U	U	+
	31	p.(Leu54Cysfs*24)	+	+	+	+	+	-	-	+	-	U	+	+	U	+	+	-	-	-	+	+	+	U	+	-	U	U	-	U	U	U	U	U	U
	T#5	p.(Ala2Profs*197)	-	U	U	U	U	U	U	U	U	U	+	+	U	+	+	-	-	+	U	U	U	U	U	U	U	U	U	U	+	U	U	U	U
	L#2	p.(Ser103Hisfs*97)	-	U	+	+	+	U	+	U	U	U	+	+	U	U	-	U	-	+	-	-	-	-	+	+	U	-	-	-	+	U	U	U	U
	L#3	p.(Gln186*)	-	U	+	+	+	U	+	U	U	U	+	+	U	U	-	U	-	-	-	-	-	-	+	+	U	-	-	-	-	U	U	U	U
	L#6	p.(Tyr121*)	-	U	+	+	+	U	+	U	U	U	+	+	U	U	+	U	-	+	-	-	-	-	+	+	U	+	+	-	-	U	U	U	U
			20%	50%	100%	100%	80%	33%	60%	58%	18%	33%	100%	100%	0%	92%	50%	9%	13%	38%	33%	23%	14%	42%	80%	57%	64%	36%	20%	29%	40%	29%	33%	17%	25%
A2	6	p.(Val226Serfs*68)	-	-	+	+	-	-	-	+	+	-	+	+	+	+	+	-	-	+	+	+	+	-	+	+	+	-	-	+	-	-	U	-	-
	8	p.(Gly268*)	-	+	+	+	+	+	+	+	+	U	+	+	-	+	+	-	-	+	+	-	+	U	+	+	+	-	-	-	+	U	U	U	-
	10	p.(Val226Glyfs*67)	+	+	+	+	+	-	-	+	+	U	+	+	+	+	+	-	+	-	+	-	-	+	+	+	+	+	-	-	+	+	-	+	+
	12	p.(Lys250Asnfs*45)	U	+	+	+	U	U	+	U	U	U	+	+	+	+	-	-	-	-	+	+	-	U	+	+	-	-	-	-	+	U	U	U	-
	27	p.(Asn238Glnfs*56)	-	-	+	+	+	-	-	-	-	U	+	+	-	+	-	+	-	+	-	-	-	U	+	+	+	-	-	-	-	U	U	U	U
	28	p.(Phe271del)	-	-	+	+	-	-	-	-	-	U	+	+	U	+	+	-	-	-	-	-	-	U	-	-	-	-	-	-	-	+	+	+	U
	30	p.(Thr270Leufs*23)	-	-	+	+	+	+	+	+	+	+	+	+	+	+	+	-	-	+	+	-	-	+	+	+	+	-	-	+	-	-	U	-	+
	32	p.(Lys229*)	-	+	+	-	+	-	-	+	-	+	+	+	+	+	-	+	-	-	-	-	-	-	+	+	-	-	-	-	-	-	U	-	-
	T#2	p.(Ile257Hisfs*37)	-	U	U	U	U	U	U	U	U	U	+	+	U	+	-	-	-	+	U	U	U	U	U	U	U	U	U	U	+	U	U	U	U
	H#1	p.(Phe243Tyrfs*50)	-	+	+	+	+	U	+	U	-	U	+	+	U	+	-	-	+	-	U	U	U	U	U	U	U	-	U	U	U	+	-	+	U
	L#1	p.(Phe271del)	-	U	+	+	+	U	+	U	U	U	+	+	U	U	+	U	-	-	-	-	-	-	U	U	U	-	-	-	-	U	U	U	U
	L#7	p.(Tyr261*)	-	U	+	+	+	U	+	U	U	U	+	+	U	U	-	U	-	-	-	-	-	-	-	-	-	-	-	-	+	U	U	U	U
			9%	56%	100%	91%	80%	29%	55%	71%	50%	67%	100%	100%	71%	100%	50%	20%	17%	42%	50%	20%	20%	33%	78%	78%	56%	9%	0%	20%	50%	50%	33%	50%	40%
B	1	p.(Arg245Pro)	-	+	+	+	+	U	U	U	U	+	+	+	U	+	-	+	+	-	+	+	-	U	U	U	U	-	-	-	U	+	+	+	U
	4	p.(Phe233del)	-	+	+	+	+	-	+	+	U	-	+	+	+	+	-	-	+	-	-	-	-	-	+	+	-	-	-	-	+	+	+	-	-
	5	p.(Phe233del)	-	+	+	+	+	-	+	+	+	+	+	+	+	+	+	+	+	-	+	+	+	+	+	+	+	+	-	-	+	+	+	-	+
	9	p.(Pro191Leu)	U	-	+	+	+	+	+	+	+	-	+	+	-	+	-	-	-	-	-	-	-	U	+	+	-	-	-	-	-	U	U	U	U
	14	p.(Phe233del)	-	+	+	+	-	U	-	U	U	U	+	+	U	+	+	-	+	-	-	-	-	U	+	+	-	-	+	-	+	+	+	+	-
	15	p.(Arg245Pro)	-	-	+	+	+	-	+	+	+	+	+	+	+	+	+	-	-	-	-	-	-	+	+	+	+	-	-	+	-	U	U	U	+
	17	p.(Ile257_Val259delinsMet)	-	+	+	+	+	-	+	+	+	U	+	+	U	+	+	-	-	+	+	+	+	-	+	+	+	+	-	+	+	U	U	U	U
	21	p.(Leu100Arg)	-	+	+	+	+	-	-	-	-	U	+	+	-	+	+	+	-	+	-	-	-	-	+	-	+	+	+	-	+	-	-	-	U
	25	p.(Ile206del)	-	-	+	+	+	-	+	-	-	U	+	+	-	+	-	-	+	+	+	-	+	-	+	+	+	-	-	-	-	-	-	-	-
	T#1	p.(Ile188Thr)	-	U	U	U	U	U	U	U	U	U	+	+	U	+	-	+	+	-	U	U	U	U	U	U	U	U	U	U	-	U	U	U	U
	T#4	p.(Phe233del)	-	U	U	U	U	U	U	U	U	U	+	+	U	+	-	-	-	+	U	U	U	U	U	U	U	U	U	U	+	U	U	U	U
	T#6	p.(Thr101_Ser103del)	-	U	U	U	U	U	U	U	U	U	+	+	U	+	-	-	-	-	U	U	U	U	U	U	U	U	U	U	+	U	U	U	U
	H#3	p.(Ile206Phe)	-	+	+	+	+	U	+	U	-	U	+	+	U	+	-	-	-	-	U	U	U	U	U	U	U	-	U	U	U	-	-	-	U
	H#4	p.(Phe233del)	-	-	+	+	+	U	+	U	+	U	+	+	U	+	+	+	+	+	U	U	U	U	U	U	U	+	U	U	+	+	+	+	U
	L#5	p.(Leu100Pro)	-	U	+	+	+	U	+	U	U	U	+	+	U	U	+	U	+	-	-	-	-	-	+	+	U	-	-	-	+	U	U	U	U
	L#8	p.(Met157Lys)	-	U	+	+	+	U	+	U	U	U	+	+	U	U	-	U	-	-	-	-	-	U	-	U	U	-	-	-	+	U	U	U	U
	L#9	p.(Ala89Pro)	-	U	+	+	+	U	+	U	U	U	+	+	U	U	+	U	+	-	+	+	-	-	U	U	U	-	+	-	-	U	U	U	U
	L#10	p.(Ile88_Ala89delinsThr)	-	U	+	+	+	U	+	U	U	U	+	+	U	U	+	U	-	-	-	-	-	+	+	+	U	+	-	-	+	U	U	U	+
	L#11	p.(Arg199Pro)	-	U	+	+	+	U	U	U	U	U	+	+	U	U	+	U	-	-	+	+	-	-	U	U	U	+	-	-	+	U	U	U	U
	O#1	p.(Arg111_Leu114del)	+	-	+	+	+	U	+	U	U	U	+	+	U	+	-	U	-	-	U	U	U	U	U	U	U	U	U	U	+	U	U	U	U
			5%	58%	100%	100%	94%	14%	87%	71%	63%	60%	100%	100%	50%	100%	50%	36%	45%	25%	43%	36%	21%	27%	100%	82%	63%	38%	21%	14%	71%	63%	63%	38%	50%
C	3	p.(Tyr74His)	+	+	+	+	+	-	-	+	-	U	+	+	-	+	-	-	+	-	-	-	-	-	+	+	U	-	-	-	-	U	U	U	+
	19	p.(Gly165_Arg169dup)	U	-	+	+	+	+	-	-	-	U	+	+	U	-	-	U	-	-	-	-	-	U	-	-	-	-	-	-	-	-	-	-	U
	20	p.(Lys97Glu)	-	+	+	+	+	-	-	+	-	U	+	+	-	+	-	-	-	-	-	-	-	-	+	-	+	-	-	-	-	U	-	-	-
	L#4	p.(Lys97Glu)	+	U	+	+	+	U	+	U	U	U	+	+	U	U	+	U	-	-	-	-	-	-	U	U	U	-	-	+	+	U	U	U	U
			67%	67%	100%	100%	100%	33%	33%	67%	0%	U	100%	100%	0%	67%	25%	0%	25%	0%	0%	0%	0%	0%	67%	33%	50%	0%	0%	25%	25%	0%	0%	0%	50%
D	H#2	p.(Glu283Argfs*45)	+	+	-	-	-	U	-	U	-	U	+	+	U	-	-	-	-	-	U	U	U	U	U	U	U	-	U	U	U	-	-	-	U
E	T#3	p.(Met1?)	-	U	U	U	U	U	U	U	U	U	+	+	U	U	+	+	-	-	U	U	U	U	U	U	U	U	U	U	+	U	U	U	U