Supplementary Table S1. Phenotype and genotype of the MAN1B1-CDG patients (summarized from *Rymen et al. [1] and **Van Scherpenzeel et al. [2])

Patients (sex, age in years)	**1 (F, 14)**	**2.1 (F, 25)**	**2.2 (M, 19)**	**3 (F, 13)**	**4 (M, 15)**
Previous patient number	***3**	***4.1**	***4.2**	***6**	****1**
Neurological involvement					
Intellectual disability	mild	mild	mild	mild	mild-moderate
Motor disability	+	+	+	+	+
Behaviour problems	-	NA	NA	autistic features	autistic features
Seizures	-	-	-	-	-
Hypotonia	+	+	+	+	+
Strabism	-	+	+	-	-
Dysmorphism					
Facial dysmorphism	+	+	+	+	+
Inverted nipples	+	-	+	+	-
Truncal obesity	+	+	+	+	+
Skin laxity	+	+	+	+	-
Skeletal and joint involvement					
Macrocephaly	-	NA	NA	-	+
Dolichocephaly	-	+	+	-	-
Short stature	+	NA	NA	-	-
Joint laxity	+	+	-	+	+
Genotype	c.1000C>T/p.R334C homozygous	c.1000C>T/p.R334C homozygous	c.1000C>T/p.R334C homozygous	c.1445+2delTGAG c.465+1460_620+527del	c.1000C>T/p.R334C homozygous

Patients (sex, age in years)	**5 (M, 14)**	**6.1 (M, 5)**	**6.2 (F, 3)**	**7 (F, 3)**	**8 (F, 17)**
Previous patient number	****3**	****5.1**	****5.2**	****4.1**	****6**
Neurological involvement					
Intellectual disability	severe	moderate-severe	moderate	moderate	moderate
Motor disability	+	+	+	+	+
Behaviour problems	autistic features/aggressivity	-	-	-	-
Seizures	-	-	-	-	-
Hypotonia	+	+	+	+	+
Strabism	+	+	-	+	+
Dysmorphism					
Facial dysmorphism	+	+	-	+	+
Inverted nipples	+	+	-	+	+
Truncal obesity	+	+	-	-	+
Skin laxity	-	-	-	-	-
Sleletal and joint involvement					
Macrocephaly	+	+	+	-	+
Dolichocephaly	-	-	-	-	-
Short stature	-	-	-	-	-
Joint laxity	-	-	-	-	-
Genotype	c.1001G>C/p.R334P c.1849C>T/p.Q617X	c.1789C>T/p.R579W c.2065G>A/p.E689K	c.1789C>T/p.R579W c.2065G>A/p.E689K	c.1863G>A/p.W621X homozygous	c.1282delA/p.1428fs*43 c.1225T>C/p.S409P

NA: not available

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