Code	Clinical classification	Ssex	Age	Max. motor ability	Climb Stairs	NdSSS	Feeding	Tongue atrophy	Ventilatory Support	age of VS	osteoesqueletics deformi	Daily life activities
P1	Intermediate	M		2 non-gravity distal movem	-		1 GT only - need aspiration	yes - pronouced	IVS /Continuos		0.1 -	NA
P2	Intermediate	F		3 non-gravity distal movem	-		1 GT only - need aspiration	yes - mild	IVS /Continuos		0.1 -	NA
P3	Typical	M		4 able to stand with support	no		2 GT only - swallow saliva	not acessed	no	-	-	compromised
P4	Typical	F		5 walk medium/long distan	unilateral support		7 OF easy-to-swallow food	not acessed	no	-	-	independent
P5	Intermediate	M		12 able to stand with support	no		3 GT, OF for fun	yes - pronouced	NIV >15h		0.1 scoliosis	compromised
P6	Intermediate	M		8 sit with support / never w	no		2 GT only - swallow saliva	yes - pronouced	IVS /Continuos		0.1 scoliosis	compromised
P7	Typical	M		9 walk medium/long distan	unilateral support		8 OF no difficult	no	no	-	-	independent
P8	Typical	M		17 walk medium/long distan	unilateral support		8 OF all kind food	no	no	-	rigid spine	independent
P9	Typical	M		17 walk short distances	unilateral support		5 OF easy-to-swallow food	no	NIVS/night-time only		11 scoliosis - surgery	compromised
P10	Childhood-onset	M		11 walk medium/long distan	independent		8 OF all kind food	no	no	-	-	independent
P11	Intermediate	F		13 sit without support/ never	no		3 GT, OF for fun	yes - pronouced	NIVS/night-time only		0.1 kyphoscoliosis	compromised
P12	Typical	M		17 sit with support / lost gait	no		6 OF easy-to-swallow food	not acessed	IVS/ Continuos		4 scoliosis - surgery	compromised
P13	Typical	F		17 walk medium/long distan	bilateral support		8 OF all kind food	no	no	-	rigid spine	compromised
P14.1	Typical	F		18 walk medium/long distan	bilateral support		5 OF easy-to-swallow food	no	NIVS />15h		12 scoliosis and thoracic def	independent
P14.2	Typical	M		21 walk medium/long distan	unilateral support		8 OF all kind food	yes - mild	NIVS/ >15h		15 scoliosis and thoracic def	independent
P15	Typical	M		13 walk short distances	bilateral support		7 OF, easy-to-swallow food	not acessed	no	-	-	independent
P16	Typical	F		13 walk medium/long distan	unilateral support		7 OF, easy-to-swallow food	no	no	-	-	independent
P17	Typical	M		15 walk medium/long distan	bilateral support		3 GTT, OF for fun	not acessed	NIVS />15h		8 -	compromised
P18	Typical	M		16 walk short distances	unilateral support		7 OF, easy-to-swallow food	yes - mild	no	-	-	independent
P19	Typical	M		18 walk medium/long distan	unilateral support		8 OF, all kind food	not acessed	no	-	rigid spine	independent
P20	Typical	M		18 walk medium/long distan	unilateral support		8 OF, all kind food	not acessed	no	-	-	independent
P21	Typical	F		25 able to stand with support	no		3 GT only,swallow saliva	yes - mild	NIVS />15h		12 scoliosis	compromised
P22.1	Typical	F		21 walk medium/long distan	bilateral support		5 OF, easy-to-swallow food	yes - mild	NIVS/ >15h		13 scoliosis and thoracic def	independent
P22.2	Typical	F		23 walk medium/long distan	unilateral support		5 OF, easy-to-swallow food	yes - mild	NIVS/night-time only		15 scoliosis and thoracic def	independent
P23	Typical	M		20 walk medium/long distan	independent		8 OF, all kind food	not acessed	no	-	-	independent
P24	Typical	F		23 walk medium/long distan	unilateral support		8 OF, all kind food	not acessed	NIVS/night-time only		15 rigid spine	independent
P25	Typical	M		27 walk short distances	no		5 OF, easy-to-swallow food	yes - pronouced	NIVS/ continuos		18 scoliosis	compromised
P26	Typical	F		31 sit with support / lost gait	no		7 OF, easy-to-swallow food	o	no	-	fingers, knee and aquileu	independent
P27	Typical	M		42 walk short distances	no		7 OF, easy-to-swallow food	no	NIVS/night-time only		15 Scoliosis - surgery	independent
P28	Childhood onset	F		44 walk medium/long distan	unilateral support		8 OF, all kind food	not acessed	NIVS/night-time only		17 rigid spine	independent
P29	Typical	F		59 walk medium/long distan	unilateral support		7 OF, easy-to-swallow food	no	no	-	scoliosis	independent
P30	Typical	F		1 non-gravity movements o	-		2 GT only, swallow saliva	yes - mild	no	used for 2 weeks	-	NA
P31	Typical	F		8 walk short distances	Bilateral support		3 GT, OF for fun	yes - pronouced	NIVS/night-time only		-	compromised

eTable 1 Clinical Findings in NM-NEB patients.

Legend: Max – Maximum, GT – Gastrostomy tube, OF – orally fed, IVS – invasive ventilatory support, NIVS – non-invasive ventilatory support,

Code	HGVSc	HGVSp	Genotype	Known variants	gnomAD (exome)	ClinVar	PMID	ACMG classification
P1	c.24735_24736del	p.Arg8245fs	HET	prev. reported	.	P	24725366	P
	c.1502_1503delinsG	p.Lys501Serfs	HET	novel	.	.	-	P
P2	c.19944G>A	p.Ser6648=	HET	prev. reported	2.82E-05	P	25205148	LP
	c.7355_7338del	p.Lys244Asnfs	HET	novel				P
P3	c.13719_13721delinsTCG	p.His4574Arg	HET	prev. reported	.	VUS	-	LP
	c.23144delA	p.Asp7715fs	HET	novel	.	.	-	P
P4	c.23140C>T	p.Arg7714*	HET	prev. reported	.		P 16917880	P
	duplication exons 95 and 96	-	HET	novel	.	.	.	LP
P5	c.7609delA	p.Lys2537fs	HET	novel	.	.	-	P
P6	c.21076C>T	p.Arg7026*	HET	prev. reported	0.00003233	.	26841830, 25205138	P
	c.24307_24308insTT	p.Tyr8103Phefs	HET	novel	.	.	-	P
P7	c.10316T>C	p.Leu3439Pro	HET	prev. reported	.	VUS	-	LP
	c.3996C>G	p.Tyr1332*	HET	prev. reported	.	P	25205138	P
P8	c.8889+1G>A	-	HET	prev. reported	.	LP	25205138	LP
	c.19102-4del	-	HET	prev. reported	.	LP	-	VUS
P9	c.5343+5G>A	-	HET	prev. reported	.	.	16917880	LP
	c.22170C>G	p.Tyr7390*	HET	prev. reported	8.04E-06	P	25205138	P
P10	c.24465_24468del	p.His8155fs	HET	novel	.	.	-	P
	c.16637G>A	p.Arg5546His	HET	prev reported	0.0009248	VUS		VUS
P11	c.21076C>T	p.Arg7026*	HET	prev. reported	0.00003233	P	26841830, 25205138	P
P12	c.24186_24189del	p.His8062fs	HET	novel	.	.	-	P
P13	c.11947_11948insAGGACTATG	p.Lys3983fs	HET	novel	.	.	-	P
	c.14878C>T	p.Arg4960*	HET	novel	6.46E-06	.	-	P
P14.1	c.24303_24304insTT	p.Leu8102fs	HOM	novel	.	.	-	P
P14.2	c.24303_24304insTT	p.Leu8102fs	HOM	novel	.	.	-	P
P15	c.9723+1G>A	-	HET	prev. reported	0	P	16199547	LP
	c.19101+5G>A	-	HET	prev. reported		P	23826317	LP
P16	*c.2647C>T*	*p.Arg883**	HET	prev. reported	0	P	25205138	P
	c.6869_6871dup	p.Ile2291insAla	HET	novel	.	.	-	LP
P17	c.9723+1G>A	-	HET	prev. reported	0	P		LP
	c.8647G>A	p.Val2883Ile	HET	prev. reported	0.00008431	Uncertain	-	LP
P18	c.13420C>T	p.Arg4474*	HET	novel	.		-	P
	Deletion (Exon 29)	-	HET	novel	-	-	-	LP
P19	c.8889+1G>A	-	HET	prev. reported	.	P	25205138	P
	c.5343+5G>A	-	HET	prev. reported	.	P	16917880	LP
P20	c.294+1G>A	-	HET	prev. reported	0	P	16199547	LP
	c.8697del	p.Lys2899Asnfs	HET	novel	.	.	-	P
P21	c.24192_24193insTCAA	p.Glu8065fs	HET	prev. reported	.	P	12207938	P
	c.7536+5G>A	-	HET	novel	.	.	-	LP
P22.1	c.23494_23495del	p.Thr7832fs	HOM	novel	.	.	-	P
P22.2	c.23494_23495del	p.Thr7832fs	HOM	novel	.	.	-	P
P23	c.24735_24736del	p.Arg8245fs	HET	prev. reported	.	P	24725366	P
	c.19621del	p.Ser6541Alafs	HET	prev. reported	.	P	-	P
P24	c.14841G>A	p.Ser4947=	HET	prev. reported	0.0000282	P	25205138	LP
	c.21418-6del	-	HET	novel	0.49	LB	-	LB
P25	c.19944G>A	p.Ser6648=	HET	prev. reported	2.82E-05	P	25205148	LP
	c.5595dup	p.Lys1866Glnfs	HET	prev. reported	.	P	-	P
P26	c.5343+5G>A	-	HET	prev. reported	.	P	16917880	LP
P27	c.5343+5G>A	-	HET	prev. reported	.	P	16917880	LP

	c.15872T>C	p.Lys5291Pro	HET	novel	.	.	-		VUS
P28	c.1674+2T>C	-	HET	prev. reported	.	P		16199547	LP
	c.8500delA	p.Lys2834fs	HET	novel	.	.	-		P
P29	c.5343+5G>A	-	HOM	prev. reported	.	P		16917880	LP
P30	*c.24219+1G>A*	-	HET	novel	-	-	-		LP
	c.7911G>C	p.Gln2637His	HET	prev. reported		1.20E-05 VUS	-		LP
P31	c.19944G>A	p.Ser6648=	HET	prev. reported		2.82E-05 P		25205148	LP
	c.1965T>G	p.Tyr655*	HET	novel	-	.			LP

eTable 2: Genetic Findings of NM-NEB patients.
Legend: HET – heterozygous, prev. – previously, HOM – homozygous, LP – likely pathogenic, P – pathogenic, VUS – variant on unknown significance.