

Pt. #	Sex	Age (y)	Diagnostic proof	Disease duration (y)	ERT status T0	Symptoms of SDB	Upright FVC (% predicted)	Δ FVC (%)	AHI (/h)	daytime pCO ₂ (mmHg)	Δ tcCO ₂ (mmHg)	tcCO ₂ max (mmHg)	Diagnosis	NIV
1	M	39	c.-32-13 T>G; c.525delT	5	-	-	105.0	15.5	10.2	36.3	3.4	44.6	OSA	-
2	M	39	c.-32-13 T>G; c.525delT	9	stopped	+	68.8	45.5	7.8	43.1	5.4	46.3	OSA, RMW	+
3	F	53	Muscle biopsy; EA [§]	28	+	+	54.9	43.8	1.6	41.0	11.4	48.9	NH	+
4	F	45	c.-32-13 T>G; del Exon 18	17	+	-	86.8	7.1	1.9	34.2	7.6	44.5	-	-
5	F	61	c.-32-13 T>G; c.307T>G	25	+	+	67.3	46.2	0.1	42.1	6.8	48.0	RMW	+
6	M	18	c.-32-13 T>G; c.1128_1129de lGGinsC	4	-	-	81.5	0	3.6	43.1	3.0	43.6	-	-
7	M	55	c.-32-13 T>G; c.701C>A	16	+	+	43.4	42.1	4.3	40.8	14.3	56.4	NH	+
8	F	45	c.-32-13 T>G; c.307T>G	33	stopped	+	57.7	19.4	1.6	36.2	2.3	47.2	RMW	+
9	F	42	c.-32-13 T>G; c.307T>G	27	+	+	48.2	47.1	11.9	39.8	5.8	46.8	OSA, RMW	+
10	M	75	c.-32-13 T>G; c.1548G>A	50	+	-	39.0	60.0	3.2	34.1	5.5	42.6	-	-

11	M	54	c.-32-13 T>G; c.525delT	19	+	+	71.6	51.7	8.5	41.1	1.0	55.0	NH	+
12	F	71	c.-32-13 T>G; c.1927G>A	16	+	+	62.6	29.4	6.3	36.5	13.7	50.8	OSA, NH	+
13	M	54	c.-32-13 T>G; c.378delTG	6	-	-	85.7	17.8	0.5	41.0	4.4	43.1	-	-
14	F	28	c.-32-13 T>G: c.2322_2323in sGGTGAGTC TGCAAACG GGGAGT	11	+	-	92.8	12.5	0.0	36.9	7.7	45.8	-	-
15	F	58	c.-32-13 T>G; c.2481+102_2 646+31del	7	+	+	44.1	53.8	0.0	40.8	3.3	45.9	RMW	+
16	F	46	c.-32-13 T>G; c.525delT	16	stopped	+	66.1	31.8	13.4	38.4	14.4	52.4	OSA, NH	+
17	F	63	c.-32-13 T>G; c.2481+102_2 646+31del	23	+	+	81.6	56.5	0.0	39.5	10.2	46.8	NH	+
18	F	42	c.-32-13 T>G; c.2481+102_2 646+31del	9	+	-	85.4	14.3	0.3	34.4	3.5	41.5	-	-
19	F	49	c.-32-13 T>G; c.1196A>T	14	-	+	64.1	66.7	6.5	53.2	37.8	85.7	OSA DH	+

20	M	71	c.-32-13 T>G; c.2481+102_2 646+31del	26	-	+	25.8	49.5	12.6	48.8	6.9	54.3	OSA DH	+
21	M	62	c.-32-13 T>G; c.525delT	4	+	+	65.2	35.7	4.1	39.0	3.2	44.5	RMW	+
22	M	78	c.-32-13 T>G; c.525delT	15	-	+	49.5	25.0	33.1	44.0	19.1	63.6	OSA, NH	+

*Table S1. Clinical data, specific GAA gene mutations, and respiratory parameters in all 22 patients with LOPD at initial evaluation (T0). The fourth column depicts the GAA genotype if available. Values relevant for the diagnosis of SDB or the clinical decision to initiate NIV are printed in bold. M, male, F, female, GAA, glucosidase alpha, EA, GAA enzyme activity, FVC, forced vital capacity, Δ FVC, positional drop of FVC, AHI, apnea hypopnea index, pCO₂, carbon dioxide tension on morning blood gas analysis, tcCO₂, transcutaneous carbon dioxide tension, NH, nocturnal hypoventilation, OSA, obstructive sleep apnea, RMW, respiratory muscle weakness (FVC<70% predicted), NIV, non-invasive ventilation, *, patient lost to follow-up, #, patient declined NIV prior to T2. §, GAA enzyme activity in this particular patient was 0,55 [mmol/h/mg Protein] at pH 4.5.*

#	Follow-up duration (months)	ERT duration prior to T0 (months)	Start or restart of ERT after T0	Upright FVC% at T0	Upright FVC% at last follow up	6-MWT distance at T0 (m)	6-MWT distance at last follow-up	Rate of FVC change (% per month)	Rate of 6-MWT distance change per month (m)	Adherence to NIV at last follow-up	Duration of NIV per 24 h at last follow-up
2	26	0	+	68.8	72.9	527	394	+0.16	-5.1	100	10.2
3	33	34	-	54.9	65.5	205	271	+0.32	+2.0	100	5.3
5	40	13	-	87.3	80.3	321	292	-0.18	-0.7	70	5.5
7	37	36	-	43.4	44.6	608	536	+0.03	-1.9	100	7.8
8	25	0	+	57.7	53.1	232	199	-0.17	-1.3	15	2.3
11	3	0	+	71.6	73.6	401	410	-0.67	+3.0	9	1.5
15	37	15	-	44.1	37.3	390	350	-0.18	-1.1	76	5.5
16	15	0	+	66.1	60.1	441	466	-0.4	+1.7	100	6.0
17	31	1	-	71.2	64.3	252	234	-0.22	-0.6	70	9.0
19	26	0	+	64.1	54.4	312	352	-0.37	+1.5	61	7.5
20	39	0	+	25.8	24.2	213	261	-0.04	+1.2	100	7.0
21	28	4	-	65.2	46.5	367	275	-0.67	-3.3	100	7.4
22	15	0	+	49.5	50.0	310	384	+0.03	+4.9	37	4.5

Table S2. ERT status, FVC, adherence to NIV, and 6-MWT results in 13 patients with LOPD who started NIV and attended follow-up visit T2. ERT, enzyme replacement therapy, FVC, forced vital capacity, 6-MWT, 6-minutes walk test, NIV, non-invasive ventilation.

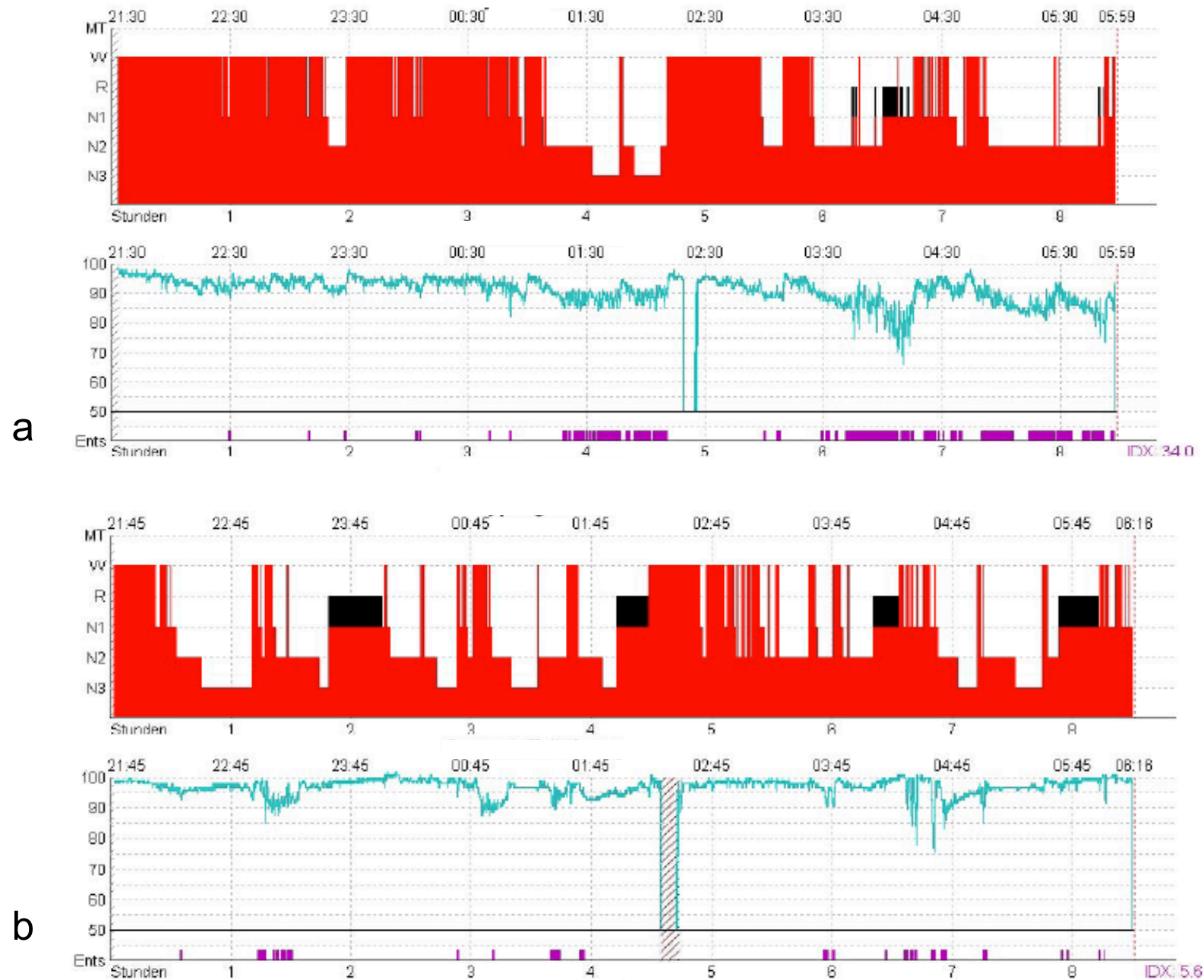


Figure S1. Hypnogram and oxygen saturation before (a) and immediately after (b) initiation of NIV.