Supplementary data

Table 1:

Admission notes from Paediatric Intensive Care Admissions amongst cohort of Congenital Myasthenic Syndrome Patients

ID	Age at Admission (years)	Genotype	Summary of Admission
4.00	1.42	COLQ	Resp acidosis. Intubated
4.00	1.25	COLQ	Resp acidosis. Intubated
4.00	0.42	COLQ	Resp ilness and seizure at home, admitted to Royal London PICU requiring ventillation
5.00	2.58	COLQ	Rapid onset of breathing difficulty. Chest infection, ventillated for two days in PICU
8.00	1.25	COL13A1	Pneumonia and severe resp failure. Admitted for 3 days
8.00	0.33	COL13A1	Admitted for chest infection and required ventillation
9.00	0.92	COLQ	May be more in the period Feb-July 2014. Sometimes NIV used rather than PICU admissions. Confident in these 3 admissions
9.00	0.25	COLQ	RSV associated (invasive vent)
9.00	0.00	COLQ	Breif period of invasive ventillation. Birmingham
14.00	2.50	COLQ	Required ventillation. Fever associated. .5yrs old
14.00	1.50	COLQ	Required ventillation. Fever associated. 1.5yrs old
14.00	0.58	COLQ	6 months old Status Epilepticus associated with resp tract infection. Last seizure Nov 2011. Pesistent recurrent resp infections reported
15.00	0.00	CHRNG	9 days ventillated
16.00	2.25	DOK7	1 week
16.00	2.00	DOK7	Throat infection -> no intubation. 48hr ventillation
16.00	1.67	DOK7	Ressuciation with Bagging in A&E after period of coughing
16.00	1.50	DOK7	Cold -> chest infection _> George's
16.00	1.33	DOK7	rsv onset
16.00	1.17	DOK7	Given Azithromyicin in a chest infection _> deteroirated, cyanosis, BiPaP full time no intubation
16.00	0.75	DOK7	Resp complicated by parainfluenza
16.00	0.33	DOK7	Repeated relapse and intubations. 5 crises over the course of a week at St. George's
16.00	0.33	DOK7	Repeated relapse and intubations. 5 crises over the course of a week at St. George's
16.00	0.33	DOK7	Repeated relapse and intubations. 5 crises over the course of a week at St. George's
16.00	0.33	DOK7	Repeated relapse and intubations. 5 crises over the course of a week at St. George's
16.00	0.33	DOK7	Repeated relapse and intubations. 5 crises over the course of a week at St. George's
16.00	0.08	DOK7	Required intubation

18.00	3.42	DOK7	Low oxygen sats, cyanosed. Transferred from secondary care
18.00	0.50	DOK7	Presented with stridor and resp difficulties
18.00	0.25	DOK7	CPAP Dependent, required intubation
19.00	2.08	CHRNE FCS	Extubated under NIV
19.00	1.08	CHRNE FCS	Resp deterioration
19.00	0.92	CHRNE FCS	Bromptom. Resp deterioration
19.00	0.25	CHRNE FCS	Cardirespiratory arrest
26.00	0.25	RAPSN	Intubutation for intercurrent infections (bronchiolitis) and weaned onto NIV
32.00	11.50	COLQ	Resp deterioration after a period of resp distress (no temp rise either). Extubated the day after and put on NIV
32.00	5.08	COLQ	Resp arrest requiring intubation and ventillation on PICU
32.00	0.50	COLQ	Similar to first episode with vomiting
32.00	0.33	COLQ	Resp crises starting with upper resp tract symptoms
34.00	17.00	RAPSN	HDU in 2013
35.00	0.83	CHAT	PICU aged 10 months for prolonged apnoea
38.00	0.00	CHAT	NICU from birth, stepped down Apr 2020

Table 2a:

Summary of Predicted Forced Vital Capacity (FVC%) and Predicted Peak Expiratory Flow (PEF%) by Congenital Myasthenic Syndrome (CMS) genotype.

Table 2b:

Raw data used to calculate Predicted Peak Expiratory Flow (PEF%) on spirometry when compared to mean predicted values of the European Respiratory Study (ERS) for PEF.

Raw data used to calculate Percentage of forced vital capacity (FVC%) on spirometry when compared to the 5th percentile of Global Lung Function Initiative (GLI) reference equations for spirometry for FVC.

ID	Genotype	FVC%	PEF%	Age at Study
10	COLQ	64.12	47.36	14.33
10	COLQ	57.27	34.59	15.42
10	COLQ	57.27	34.59	15.42
10	COLQ	57.69	54.39	15.92
10	COLQ	57.69	54.39	15.92
11	COL13A1	67.94	61.52	13.00
11	COL13A1	69.87	63.05	13.58
11	COL13A1	59.23	75.32	14.75
11	COL13A1	68.21	83.90	15.75
31	RAPSN	58.34	64.00	14.17

31	RAPSN	47.14	51.65	15.33
31	RAPSN	41.23	67.97	16.75
6	COLQ	105.33	79.25	9.50
6	COLQ	105.33	79.25	9.50
6	COLQ	94.37	96.62	10.33
6	COLQ	94.37	96.62	10.33
6	COLQ	94.37	96.62	10.33
6	COLQ	94.37	96.62	10.33
6	COLQ	86.38	78.80	11.42
6	COLQ	86.38	78.80	11.42
6	COLQ	86.38	78.80	11.42
6	COLQ	86.38	78.80	11.42
6	COLQ	83.83	90.99	12.83
12	CHRNA1	103.33	91.86	7.83
12	CHRNA1	88.57	74.64	9.25
12	CHRNA1	91.30	59.22	9.25
17	DOK7	72.62	39.44	11.33
17	DOK7	56.41	46.44	13.08
17	DOK7	56.41	46.44	13.08
17	DOK7	55.15	43.93	13.50
17	DOK7	54.51	45.37	15.25
20	DOK7	111.20	110.01	15.67
20	DOK7	112.15	99.69	16.83
21	DOK7	81.01	92.56	8.33
21	DOK7	82.83	88.70	8.33
21	DOK7	93.90	104.89	10.33
21	DOK7	83.51	108.77	14.67
21	DOK7	88.23	115.79	16.33
21	DOK7	81.08	105.70	17.17
25	COLQ	#NULL!	#NULL!	11.46
25	COLQ	#NULL!	#NULL!	12.57
25	COLQ	75.00	#NULL!	14.58
29	RAPSN	91.12	102.60	14.83
29	RAPSN	96.14	116.77	15.33
29	RAPSN	95.16	113.80	15.67
29	RAPSN	93.44	101.17	8.50
29	RAPSN	91.34	107.97	10.75
29	RAPSN	91.34	107.97	10.75
29	RAPSN	91.34	107.97	10.75
29	RAPSN	91.34	107.97	10.75
29	RAPSN	95.04	94.47	11.25
29	RAPSN	95.04	94.47	11.25
29	RAPSN	95.04	94.47	11.25
29	RAPSN	95.04	94.47	11.25
29	RAPSN	95.50	105.48	11.25
29	RAPSN	95.50	105.48	11.25

29	RAPSN	95.50	105.48	11.25
29	RAPSN	95.50	105.48	11.25
29	RAPSN	100.76	104.26	11.67
29	RAPSN	100.76	104.26	11.67
29	RAPSN	97.81	105.02	12.25
29	RAPSN	97.81	105.02	12.25
29	RAPSN	94.24	102.24	13.25
29	RAPSN	90.26	115.83	13.83
31	RAPSN	39.97	72.07	17.17
32	COLQ	72.64	49.98	14.58
32	COLQ	76.49	46.73	15.08
32	COLQ	76.49	46.73	15.08
32	COLQ	80.94	49.18	15.50
32	COLQ	80.94	49.18	15.50
32	COLQ	82.53	48.28	15.92
32	COLQ	82.53	48.28	15.92
32	COLQ	76.45	47.57	16.50
33	DOK7	57.61	42.11	17.17
33	DOK7	49.83	52.83	17.33
33	DOK7	50.78	37.45	13.67
33	DOK7	62.71	35.52	14.67
33	DOK7	53.80	40.36	15.58
33	DOK7	55.89	43.47	16.83
4	COLQ	#NULL!	89.14	3.92
4	COLQ	#NULL!	88.16	4.67
4	COLQ	79.44	96.92	5.75
5	COLQ	71.30	59.58	5.25
5	COLQ	86.26	71.24	5.75
5	COLQ	97.48	69.51	6.50
5	COLQ	92.23	70.19	7.58
5	COLQ	92.23	70.19	7.58
14	COLQ	#NULL!	70.01	4.25
14	COLQ	#NULL!	98.37	4.42
14	COLQ	71.60	68.65	5.25
14	COLQ	89.31	76.73	6.67
14	COLQ	75.83	65.32	7.17
14	COLQ	83.92	87.71	7.92
14	COLQ	85.49	85.67	9.00
18	DOK7	35.28	35.76	10.08
19	CHRNE FCS	#NULL!	#NULL!	3.75
19	CHRNE FCS	74.08	73.22	6.67
19	CHRNE FCS	60.80	62.82	7.42
19	CHRNE FCS	65.29	58.61	8.50
19	CHRNE FCS	56.09	81.89	10.17
19	CHRNE FCS	67.70	87.80	10.50
19	CHRNE FCS	67.70	87.80	10.50

23	DOK7	70.24	56.29	5.67
23	DOK7	86.09	58.43	7.67
23	DOK7	80.38	47.66	11.58
23	DOK7	80.38	47.66	11.58
23	DOK7	64.95	50.79	16.08
28	DOK7	62.90	45.98	8.42
28	DOK7	70.65	51.78	8.42
28	DOK7	88.19	70.52	8.67
28	DOK7	90.36	65.52	8.67
28	DOK7	91.81	55.96	8.67
28	DOK7	95.43	31.52	9.25
28	DOK7	97.17	45.78	9.67
28	DOK7	91.89	63.73	9.75
28	DOK7	93.08	40.25	10.17
28	DOK7	95.16	47.41	10.17
28	DOK7	88.68	40.71	10.67
28	DOK7	93.27	42.40	10.67
28	DOK7	97.11	52.67	10.67
28	DOK7	86.34	56.22	14.25
28	DOK7	78.91	57.93	15.50
28	DOK7	78.91	57.93	15.50
28	DOK7	81.62	65.26	16.17
28	DOK7	81.62	65.26	16.17
28	DOK7	84.94	61.35	16.17
28	DOK7	84.94	61.35	16.17
13	CHRNE	64.08	48.34	9.75
13	CHRNE	64.08	48.34	9.75
13	CHRNE	70.59	57.50	10.17
13	CHRNE	70.59	57.50	10.17
13	CHRNE	70.59	57.50	10.17
13	CHRNE	70.59	57.50	10.17
13	CHRNE	74.19	54.24	11.00
13	CHRNE	74.19	54.24	11.00
13	CHRNE	67.42	56.69	11.67
13	CHRNE	76.91	73.14	12.33
13	CHRNE	70.17	80.66	14.17
16	DOK7	#NULL!	53.10	5.75
22	CHRNE	92.52	57.59	7.58
22	CHRNE	86.65	74.93	10.08
22	CHRNE	81.91	68.97	11.25
22	CHRNE	81.06	68.78	12.42
24	COLQ	86.10	53.77	11.42
26	RAPSN	#NULL!	59.90	6.67
26	RAPSN	91.68	78.70	7.25
26	RAPSN	98.26	76.17	8.25
26	RAPSN	82.10	63.97	9.33

26	RAPSN	87.59	88.13	13.08
26	RAPSN	87.59	88.13	13.08
26	RAPSN	92.63	95.04	14.00
26	RAPSN	92.63	95.04	14.00
26	RAPSN	90.25	84.15	12.25
26	RAPSN	90.25	84.15	12.25
27	RAPSN	94.84	65.70	7.83
27	RAPSN	88.64	71.95	12.00
27	RAPSN	88.64	71.95	12.00
27	RAPSN	84.47	72.55	13.92
27	RAPSN	81.60	75.00	14.92
27	RAPSN	82.78	80.01	15.42
30	CHRNA SCS	#NULL!	69.89	4.50
30	CHRNA SCS	83.14	76.34	7.83
30	CHRNA SCS	83.65	63.78	8.75
30	CHRNA SCS	85.11	78.71	9.67

Genotype	ID	Height (m)	Gender	Age (years)	FEV1	FVC	Reference FVC at GLI	PEF	Reference PEF at ERS	FVC%	PEF%
COLQ	10	1.47	Female	14.33	1.429	1.526	2.38	154.86	327.00	64.12	47.36
COLQ	10	1.47	Female	15.42	1.155	1.363	2.38	113.10	327.00	57.27	34.59
COLQ	10	1.47	Female	15.42	1.155	1.363	2.38	113.10	327.00	57.27	34.59
COLQ	10	1.47	Female	15.92	1.330	1.373	2.38	177.84	327.00	57.69	54.39
COLQ	10	1.47	Female	15.92	1.330	1.373	2.38	177.84	327.00	57.69	54.39
COL13A2	11	1.39	Male	13.00	1.399	1.617	2.38	193.80	315.00	67.94	61.52
COL13A2	11	1.41	Male	13.58	1.289	1.663	2.38	198.60	315.00	69.87	63.05
COL13A2	11	1.42	Male	14.75	1.426	1.617	2.73	237.26	315.00	59.23	75.32
COL13A2	11	1.45	Male	15.75	1.501	1.862	2.73	287.76	343.00	68.21	83.90
RPSN	31	1.41	Male	14.17	1.475	1.511	2.59	201.60	315.00	58.34	64.00
RPSN	31	1.55	Male	15.33	1.450	1.565	3.32	205.58	398.00	47.14	51.65
RPSN	31	1.49	Male	16.75	1.265	1.369	3.32	252.15	371.00	41.23	67.97
COLQ	6	1.39	Female	9.50	1.924	2.212	2.10	239.34	302.00	105.33	79.25
COLQ	6	1.39	Female	9.50	1.924	2.212	2.10	239.34	302.00	105.33	79.25
COLQ	6	1.44	Female	10.33	2.008	2.246	2.38	315.96	327.00	94.37	96.62
COLQ	6	1.44	Female	10.33	2.008	2.246	2.38	315.96	327.00	94.37	96.62
COLQ	6	1.44	Female	10.33	2.008	2.246	2.38	315.96	327.00	94.37	96.62
COLQ	6	1.44	Female	10.33	2.008	2.246	2.38	315.96	327.00	94.37	96.62
COLQ	6	1.51	Female	11.42	1.943	2.289	2.65	276.60	351.00	86.38	78.80
COLQ	6	1.51	Female	11.42	1.943	2.289	2.65	276.60	351.00	86.38	78.80
COLQ	6	1.51	Female	11.42	1.943	2.289	2.65	276.60	351.00	86.38	78.80
COLQ	6	1.51	Female	11.42	1.943	2.289	2.65	276.60	351.00	86.38	78.80
COLQ	6	1.59	Female	12.83	2.639	2.909	3.47	363.06	399.00	83.83	90.99
CHRNA1	12	1.16	Female	7.83	1.254	1.426	1.38	166.26	181.00	103.33	91.86
CHRNA1	12	1.23	Female	9.25	1.308	1.426	1.61	171.66	230.00	88.57	74.64
CHRNA1	12	1.23	Female	9.25	1.238	1.470	1.61	136.20	230.00	91.30	59.22
DOK7	17	1.38	Female	11.33	1.402	1.525	2.10	119.10	302.00	72.62	39.44
DOK7	17	1.49	Female	13.08	1.653	1.743	3.09	163.01	351.00	56.41	46.44
DOK7	17	1.49	Female	13.08	1.653	1.743	3.09	163.01	351.00	56.41	46.44
DOK7	17	1.52	Female	13.50	1.600	1.704	3.09	154.18	351.00	55.15	43.93
DOK7	17	1.66	Female	15.25	1.915	2.115	3.88	192.36	424.00	54.51	45.37

DOK7	20	1.62	Male	15.67	3.989	4.181	3.76	468.66	426.00	111.20	110.01
DOK7	20	1.62	Male	16.83	3.808	4.217	3.76	424.70	426.00	112.15	99.69
DOK7	21	1.23	Female	8.33	1.106	1.118	1.38	212.88	230.00	81.01	92.56
DOK7	21	1.23	Female	8.33	1.125	1.143	1.38	204.00	230.00	82.83	88.70
DOK7	21	1.33	Female	10.33	1.382	1.493	1.59	291.60	278.00	93.90	104.89
DOK7	21	1.50	Female	14.67	2.053	2.213	2.65	381.78	351.00	83.51	108.77
DOK7	21	1.51	Female	16.33	2.103	2.338	2.65	406.44	351.00	88.23	115.79
DOK7	21	1.57	Female	17.17	2.168	2.408	2.97	396.36	375.00	81.08	105.70
COLQ	25	#NULL!	Male	11.46	1.840	2.250	#NULL!	#NULL!	#NULL!	#NULL!	#NULL!
COLQ	25	#NULL!	Male	12.57	1.770	2.040	#NULL!	#NULL!	#NULL!	#NULL!	#NULL!
COLQ	25	1.62	Male	14.58	#NULL!	2.820	3.76	#NULL!	426.00	75.00	#NULL!
RPSN	29	1.59	Female	14.83	3.105	3.162	3.47	409.37	399.00	91.12	102.60
RPSN	29	1.60	Female	15.33	3.219	3.336	3.47	465.90	399.00	96.14	116.77
RPSN	29	1.60	Female	15.67	3.273	3.302	3.47	454.08	399.00	95.16	113.80
RPSN	29	1.28	Female	8.50	1.531	1.738	1.86	256.98	254.00	93.44	101.17
RPSN	29	1.42	Female	10.75	2.024	2.174	2.38	326.08	302.00	91.34	107.97
RPSN	29	1.42	Female	10.75	2.024	2.174	2.38	326.08	302.00	91.34	107.97
RPSN	29	1.42	Female	10.75	2.024	2.174	2.38	326.08	302.00	91.34	107.97
RPSN	29	1.42	Female	10.75	2.024	2.174	2.38	326.08	302.00	91.34	107.97
RPSN	29	1.44	Female	11.25	2.038	2.262	2.38	308.91	327.00	95.04	94.47
RPSN	29	1.44	Female	11.25	2.038	2.262	2.38	308.91	327.00	95.04	94.47
RPSN	29	1.44	Female	11.25	2.038	2.262	2.38	308.91	327.00	95.04	94.47
RPSN	29	1.44	Female	11.25	2.038	2.262	2.38	308.91	327.00	95.04	94.47
RPSN	29	1.44	Female	11.25	2.114	2.273	2.38	344.92	327.00	95.50	105.48
RPSN	29	1.44	Female	11.25	2.114	2.273	2.38	344.92	327.00	95.50	105.48
RPSN	29	1.44	Female	11.25	2.114	2.273	2.38	344.92	327.00	95.50	105.48
RPSN	29	1.44	Female	11.25	2.114	2.273	2.38	344.92	327.00	95.50	105.48
RPSN	29	1.47	Female	11.67	2.179	2.398	2.38	340.92	327.00	100.76	104.26
RPSN	29	1.47	Female	11.67	2.179	2.398	2.38	340.92	327.00	100.76	104.26
RPSN	29	1.51	Female	12.25	2.405	2.592	2.65	368.63	351.00	97.81	105.02

RPSN	29	1.51	Female	12.25	2.405	2.592	2.65	368.63	351.00	97.81	105.02
RPSN	29	1.56	Female	13.25	2.801	2.912	3.09	383.40	375.00	94.24	102.24
RPSN	29	1.57	Female	13.83	2.970	3.132	3.47	434.37	375.00	90.26	115.83
RPSN	31	1.48	Male	17.17	1.266	1.327	3.32	267.37	371.00	39.97	72.07
COLQ	32	1.51	Female	14.58	1.461	1.925	2.65	175.44	351.00	72.64	49.98
COLQ	32	1.51	Female	15.08	1.560	2.027	2.65	164.01	351.00	76.49	46.73
COLQ	32	1.51	Female	15.08	1.560	2.027	2.65	164.01	351.00	76.49	46.73
COLQ	32	1.52	Female	15.50	1.826	2.145	2.65	172.63	351.00	80.94	49.18
COLQ	32	1.52	Female	15.50	1.826	2.145	2.65	172.63	351.00	80.94	49.18
COLQ	32	1.52	Female	15.92	1.888	2.187	2.65	169.45	351.00	82.53	48.28
COLQ	32	1.52	Female	15.92	1.888	2.187	2.65	169.45	351.00	82.53	48.28
COLQ	32	1.52	Female	16.50	1.612	2.026	2.65	166.98	351.00	76.45	47.57
DOK7	33	1.64	Male	17.17	1.717	2.166	3.76	191.20	454.00	57.61	42.11
DOK7	33	1.72	Male	17.33	1.805	2.327	4.67	254.64	482.00	49.83	52.83
DOK7	33	1.51	Male	13.67	1.564	1.686	3.32	138.95	371.00	50.78	37.45
DOK7	33	1.56	Male	14.67	1.776	2.082	3.32	141.35	398.00	62.71	35.52
DOK7	33	1.61	Male	15.58	1.816	2.023	3.76	171.95	426.00	53.80	40.36
DOK7	33	1.72	Male	16.83	1.864	2.364	4.23	209.52	482.00	55.89	43.47
COLQ	4	1.07	Male	3.92	0.813	0.849	#NULL!	106.08	119.00	#NULL!	89.14
COLQ	4	1.12	Male	4.67	0.870	0.897	#NULL!	129.60	147.00	#NULL!	88.16
COLQ	4	1.20	Male	5.75	1.133	1.144	1.44	196.74	203.00	79.44	96.92
COLQ	5	1.08	Female	5.25	0.820	0.820	1.15	93.54	157.00	71.30	59.58
COLQ	5	1.11	Female	5.75	0.943	0.992	1.15	111.84	157.00	86.26	71.24
COLQ	5	1.16	Female	6.50	1.048	1.121	1.15	125.82	181.00	97.48	69.51
COLQ	5	1.22	Female	7.58	1.286	1.365	1.48	143.88	205.00	92.23	70.19
COLQ	5	1.22	Female	7.58	1.286	1.365	1.48	143.88	205.00	92.23	70.19
COLQ	14	1.07	Male	4.25	0.618	0.638	#NULL!	83.31	119.00	#NULL!	70.01
COLQ	14	1.09	Male	4.42	0.640	0.662	#NULL!	144.60	147.00	#NULL!	98.37
COLQ	14	1.15	Male	5.25	0.899	1.031	1.44	120.14	175.00	71.60	68.65
COLQ	14	1.24	Male	6.67	1.198	1.286	1.44	177.24	231.00	89.31	76.73

COLQ	14	1.28	Male	7.17	1.248	1.274	1.68	150.90	231.00	75.83	65.32
COLQ	14	1.31	Male	7.92	1.451	1.519	1.81	227.16	259.00	83.92	87.71
COLQ	14	1.37	Male	9.00	1.693	1.761	2.06	245.88	287.00	85.49	85.67
DOK7	18	1.32	Female	10.08	0.514	0.561	1.59	90.84	254.00	35.28	35.76
FCS	19	0.93	Female	3.75	0.368	0.377	#NULL!	77.26	#NULL!	#NULL!	#NULL!
FCS	19	1.13	Female	6.67	0.858	0.926	1.25	132.52	181.00	74.08	73.22
FCS	19	1.13	Female	7.42	0.683	0.760	1.25	113.70	181.00	60.80	62.82
FCS	19	1.16	Female	8.50	0.799	0.901	1.38	106.08	181.00	65.29	58.61
FCS	19	1.22	Female	10.17	0.880	0.903	1.61	167.88	205.00	56.09	81.89
FCS	19	1.22	Female	10.50	1.069	1.090	1.61	180.00	205.00	67.70	87.80
FCS	19	1.22	Female	10.50	1.069	1.090	1.61	180.00	205.00	67.70	87.80
DOK7	23	1.08	Female	5.67	0.818	0.878	1.25	88.38	157.00	70.24	56.29
DOK7	23	1.22	Female	7.67	0.997	1.188	1.38	119.77	205.00	86.09	58.43
DOK7	23	1.41	Female	11.58	1.361	1.688	2.10	143.93	302.00	80.38	47.66
DOK7	23	1.41	Female	11.58	1.361	1.688	2.10	143.93	302.00	80.38	47.66
DOK7	23	1.65	Female	16.08	1.944	2.520	3.88	215.34	424.00	64.95	50.79
DOK7	28	1.21	Female	8.42	0.643	0.868	1.38	94.26	205.00	62.90	45.98
DOK7	28	1.21	Female	8.42	0.718	0.975	1.38	106.14	205.00	70.65	51.78
DOK7	28	1.20	Female	8.67	1.038	1.217	1.38	144.58	205.00	88.19	70.52
DOK7	28	1.20	Female	8.67	1.056	1.247	1.38	134.32	205.00	90.36	65.52
DOK7	28	1.22	Female	8.67	1.046	1.267	1.38	114.73	205.00	91.81	55.96
DOK7	28	1.24	Female	9.25	1.019	1.317	1.38	72.50	230.00	95.43	31.52
DOK7	28	1.26	Female	9.67	0.926	1.341	1.38	105.29	230.00	97.17	45.78
DOK7	28	1.27	Female	9.75	1.258	1.461	1.59	146.57	230.00	91.89	63.73
DOK7	28	1.28	Female	10.17	1.081	1.480	1.59	102.24	254.00	93.08	40.25
DOK7	28	1.28	Female	10.17	1.186	1.513	1.59	120.41	254.00	95.16	47.41
DOK7	28	1.28	Female	10.67	1.067	1.410	1.59	103.40	254.00	88.68	40.71
DOK7	28	1.30	Female	10.67	1.137	1.483	1.59	107.71	254.00	93.27	42.40
DOK7	28	1.30	Female	10.67	1.324	1.544	1.59	133.78	254.00	97.11	52.67
DOK7	28	1.48	Female	14.25	1.644	2.055	2.38	197.34	351.00	86.34	56.22

DOK7	28	1.49	Female	15.50	1.691	2.091	2.65	203.34	351.00	78.91	57.93
DOK7	28	1.49	Female	15.50	1.691	2.091	2.65	203.34	351.00	78.91	57.93
DOK7	28	1.50	Female	16.17	1.918	2.163	2.65	229.08	351.00	81.62	65.26
DOK7	28	1.50	Female	16.17	1.918	2.163	2.65	229.08	351.00	81.62	65.26
DOK7	28	1.50	Female	16.17	1.941	2.251	2.65	215.34	351.00	84.94	61.35
DOK7	28	1.50	Female	16.17	1.941	2.251	2.65	215.34	351.00	84.94	61.35
CHRNE	13	1.44	Female	9.75	1.456	1.525	2.38	158.07	327.00	64.08	48.34
CHRNE	13	1.44	Female	9.75	1.456	1.525	2.38	158.07	327.00	64.08	48.34
CHRNE	13	1.47	Female	10.17	1.589	1.680	2.38	188.02	327.00	70.59	57.50
CHRNE	13	1.47	Female	10.17	1.589	1.680	2.38	188.02	327.00	70.59	57.50
CHRNE	13	1.47	Female	10.17	1.589	1.680	2.38	188.02	327.00	70.59	57.50
CHRNE	13	1.47	Female	10.17	1.589	1.680	2.38	188.02	327.00	70.59	57.50
CHRNE	13	1.54	Female	11.00	1.845	1.966	2.65	203.40	375.00	74.19	54.24
CHRNE	13	1.54	Female	11.00	1.845	1.966	2.65	203.40	375.00	74.19	54.24
CHRNE	13	1.58	Female	11.67	1.927	2.009	2.98	226.20	399.00	67.42	56.69
CHRNE	13	1.59	Female	12.33	2.223	2.292	2.98	291.84	399.00	76.91	73.14
CHRNE	13	1.62	Female	14.17	2.317	2.435	3.47	321.84	399.00	70.17	80.66
DOK7	16	1.12	Male	5.75	0.762	0.874	#NULL!	78.06	147.00	#NULL!	53.10
CHRNE	22	1.26	Male	7.58	1.311	1.434	1.55	133.04	231.00	92.52	57.59
CHRNE	22	1.39	Male	10.08	1.649	1.785	2.06	236.02	315.00	86.65	74.93
CHRNE	22	1.44	Male	11.25	1.714	1.933	2.36	236.58	343.00	81.91	68.97
CHRNE	22	1.50	Male	12.42	1.856	2.140	2.64	255.18	371.00	81.06	68.78
COLQ	24	1.49	Male	11.42	2.290	2.471	2.87	199.50	371.00	86.10	53.77
RPSN	26	1.13	Female	6.67	0.955	1.021	#NULL!	108.41	181.00	#NULL!	59.90
RPSN	26	1.16	Female	7.25	1.059	1.146	1.25	142.45	181.00	91.68	78.70
RPSN	26	1.22	Female	8.25	1.227	1.356	1.38	156.14	205.00	98.26	76.17
RPSN	26	1.27	Female	9.33	1.362	1.527	1.86	147.12	230.00	82.10	63.97
RPSN	26	1.47	Female	13.08	2.191	2.435	2.78	288.18	327.00	87.59	88.13
RPSN	26	1.47	Female	13.08	2.191	2.435	2.78	288.18	327.00	87.59	88.13
RPSN	26	1.48	Female	14.00	2.132	2.575	2.78	333.60	351.00	92.63	95.04

RPSN	26	1.48	Female	14.00	2.132	2.575	2.78	333.60	351.00	92.63	95.04
RPSN	26	1.44	Female	12.25	1.939	2.148	2.38	275.16	327.00	90.25	84.15
RPSN	26	1.44	Female	12.25	1.939	2.148	2.38	275.16	327.00	90.25	84.15
RPSN	27	1.26	Female	7.83	1.290	1.527	1.61	151.11	230.00	94.84	65.70
RPSN	27	1.50	Female	12.00	1.986	2.349	2.65	252.53	351.00	88.64	71.95
RPSN	27	1.50	Female	12.00	1.986	2.349	2.65	252.53	351.00	88.64	71.95
RPSN	27	1.63	Female	13.92	2.375	2.931	3.47	307.62	424.00	84.47	72.55
RPSN	27	1.65	Female	14.92	2.756	3.166	3.88	318.00	424.00	81.60	75.00
RPSN	27	1.66	Female	15.42	2.747	3.212	3.88	339.24	424.00	82.78	80.01
SCS	30	1.03	Male	4.50	0.393	0.393	#NULL!	63.60	91.00	#NULL!	69.89
SCS	30	1.24	Male	7.83	1.241	1.405	1.69	176.34	231.00	83.14	76.34
SCS	30	1.30	Male	8.75	1.341	1.648	1.97	165.18	259.00	83.65	63.78
SCS	30	1.36	Male	9.67	1.569	1.898	2.23	225.90	287.00	85.11	78.71

Table 3:

Cardiorespiratory sleep study data from the cohort of Congenital Myasthenic Syndrome patients.

- tCO2 – transcutaneous carbon dioxide
- ODI – Oxygen Desaturation Index
- AHI – Apnoea/hypopnoea index

ID	Age at Study	Genotype	Mean tCO2	ODI	AHI
1.00	1.17	CHRNE	39	4	1.6
4.00	1.92	COLQ	39	1	0.5
4.00	3.17	COLQ	43	0	0.1
5.00	3.75	COLQ	41	15	11.1
5.00	4.92	COLQ	42	3	0.8
6.00	9.92	COLQ	41	3	1.8
6.00	10.92	COLQ	45	1	1.1

6.00	12.25	COLQ	45	2	1.2
6.00	13.20	COLQ	45	2	1.0
8.00	5.25	COL13A1	40	21	7.4
10.00	12.08	COLQ	45	6	2.4
10.00	13.08	COLQ	48	8	6.7
10.00	15.92	COLQ	42	29	8.7
13.00	8.58	CHRNE	40	0	0.0
14.00	3.25	COLQ	43	2	1.0
14.00	5.08	COLQ	42	1	0.7
14.00	6.67	COLQ	45	0	0.1
14.00	8.75	COLQ	45	3	0.0
15.00	0.17	CHRNG	75	26	8.8
15.00	0.17	CHRNG	66	9	3.9
15.00	0.25	CHRNG	62	45	8.8
15.00	0.33	CHRNG	61	6	3.1
15.00	0.83	CHRNG	62	8	2.2
15.00	0.92	CHRNG	64	23	6.9
15.00	0.92	CHRNG	55	30	10.5
15.00	1.17	CHRNG	58	12	2.4
15.00	1.83	CHRNG	51	13	2.1
15.00	2.25	CHRNG	53	11	2.4
15.00	2.58	CHRNG	47	18	1.2
15.00	3.08	CHRNG	59	17	5.4
15.00	3.58	CHRNG	52	6	4.6
15.00	4.08	CHRNG	56	8	3.9
15.00	4.67	CHRNG	54	9	2.2
15.00	5.75	CHRNG	55	7	2.7
15.00	6.67	CHRNG	50	10	2.9
15.00	8.06	CHRNG	50	5	1.6
15.00	8.06	CHRNG	51	6	1.9

15.00	8.44	CHRNG	51	3	0.1
15.00	8.44	CHRNG	54	10	2.7
17.00	15.33	DOK7	51	13	10.8
17.00	15.42	DOK7	48	3	1.5
17.00	15.42	DOK7	50	4	0.9
17.00	15.67	DOK7	38	8	4.6
17.00	16.42	DOK7	37	0	0.0
18.00	0.75	DOK7	47	3	0.7
18.00	1.17	DOK7	46	19	7.5
18.00	1.50	DOK7	46	21	10.7
18.00	1.75	DOK7	50	20	12.5
18.00	1.83	DOK7	46	32	2.4
18.00	2.25	DOK7	45	3	0.4
18.00	2.75	DOK7	47	8	0.0
18.00	4.25	DOK7	51	27	1.5
18.00	5.00	DOK7	35	18	0.2
18.00	5.75	DOK7	77	59	0.1
18.00	6.83	DOK7	54	1	0.4
18.00	7.33	DOK7	69	17	0.0
18.00	7.58	DOK7	56	13	#NULL!
18.00	7.58	DOK7	41	0	0.0
18.00	7.92	DOK7	55	3	0.0
18.00	8.83	DOK7	50	28	13.1
18.00	9.58	DOK7	44	8	0.0
21.00	10.33	DOK7	50	5	4.1
21.00	12.25	DOK7	49	6	1.3
21.00	13.08	DOK7	46	5	2.3
21.00	14.42	DOK7	40	5	1.4
21.00	15.83	DOK7	48	3	2.4
23.00	7.83	DOK7	43	0	0.3

23.00	13.50	DOK7	45	0	0.3
28.00	9.00	DOK7	46	3	2.1
28.00	10.00	DOK7	47	3	2.3
28.00	11.17	DOK7	49	7	2.3
28.00	12.58	DOK7	44	3	1.7
29.00	9.92	RPSN	46	7	0.8
29.00	11.00	RPSN	43	0	0.0
29.00	12.08	RPSN	48	0	0.0
29.00	13.08	RPSN	46	0	0.1
29.00	14.17	RPSN	38	0	0.0
29.00	15.08	RPSN	40	0	0.0
29.00	16.42	RPSN	40	0	0.0
30.00	10.75	SCS	47	4	2.2
30.00	12.08	SCS	45	5	3.3
31.00	15.67	RPSN	50	2	1.1
31.00	16.33	RPSN	36	1	0.3
31.00	17.42	RPSN	50	0	0.0
32.00	11.50	COLQ	49	3	0.3
32.00	12.00	COLQ	46	1	0.1
32.00	13.08	COLQ	46	5	3.3
32.00	14.08	COLQ	#NULL!	1	0.0
32.00	14.75	COLQ	49	0	0.0
32.00	15.75	COLQ	52	2	0.0
32.00	16.83	COLQ	39	0	0.0
33.00	16.83	DOK7	57	11	8.4
33.00	17.08	DOK7	54	11	3.6
33.00	17.08	DOK7	54	8	2.5
33.00	17.33	DOK7	51	1	1.4
33.00	17.75	DOK7	49	1	0.6
33.00	18.08	DOK7	48	4	2.8