

Supplementary table 4 Comparisons of clinical characteristics between cases from publications and our study

	Group 1(n=43)	Group2 (n=57)	Group 3 (n=16)	Total (n=116)	p1* value	p2# value
Gender (n)	37	57	16	110	-	-
Male (n/total, %)	29/37, 78.4	23/57, 40.4	8/16, 50.0	60/110, 54.5	>0.05	>0.05
Female (n/total, %)	8/37, 21.6	34/57, 59.6	8/16, 50.0	50/110, 45.5	>0.05	>0.05
Obesity (n/total, %)	43/43, 100	53/57, 93.0	16/16, 100	112/116, 96.6	>0.05	>0.05
Hypoventilation (n/total, %)	41/43, 95.3	47/57, 82.5	15/16, 93.8	103/116, 88.8	>0.05	>0.05
Nocturnal hypoventilation (n/total, %)	NA	42/57, 73.7	15/16, 93.8	57/73, 78.1	>0.05	>0.05
OSAS (n/total, %)	15/41, 36.6	30/47, 63.8	9/13, 69.2	54/101, 53.5	>0.05	>0.05
Daytime hypoventilation (n/total, %)	NA	10/47, 21.3	5/16, 31.2	15/64, 23.4	>0.05	>0.05
Cardiorespiratory arrest (n/total, %)	2/41, 4.9	3/47, 6.4	3/16, 13.3	8/104, 7.7	>0.05	>0.05
Hypothalamic dysfunction (n/total, %)	43/43, 100	47/57, 82.5	15/16, 93.8	105/116, 90.5	>0.05	>0.05
Hyperprolactinemia (n/total, %)	22/43, 51.2	27/47, 57.4	9/16, 56.2	58/106, 54.7	>0.05	>0.05
Hypernatremia (n/total, %)	19/43, 44.2	21/47, 44.7	9/16, 56.2	49/106, 46.2	>0.05	>0.05
Polyuria (n/total, %)	9/43, 20.9	18/47, 38.3	8/16, 50.0	35/106, 33.0	>0.05	>0.05
Hypothyroidism (n/total, %)	12/43, 27.9	20/47, 42.6	6/16, 37.5	38/106, 35.8	>0.05	>0.05
Low IGF-1 level (n/total, %)	12/43, 27.9	18/47, 38.3	8/16, 50.0	38/106, 35.8	>0.05	>0.05
Adrenocortical insufficiency (n/total, %)	NA	9/47, 19.1	1/16, 6.3	10/63, 15.9	>0.05	>0.05
Precocious puberty (n/total, %)	5/43, 11.6	9/47, 19.1	0/16, 0	14/106, 13.2	>0.05	>0.05
Delayed puberty (n/total, %)	4/43, 9.3	3/47, 6.4	1/16, 6.3	8/106, 7.5	>0.05	>0.05
Autonomic dysregulation (n/total, %)	35/43, 81.4	47/57, 82.5	16/16, 100	98/116, 84.5	>0.05	>0.05
Thermal dysregulation (n/total, %)	23/35, 65.7	8/47, 17.0	11/16, 68.8	42/98, 42.9	<0.01	<0.01

Cardiovascular manifestations (arrhythmias or blood pressure dysregulation) (n/total, %)	NA	6/47, 12.8	13/16, 81.2	19/63, 30.2	-	<0.01
Excessive sweating (n/total, %)	10/35, 28.6	5/47, 10.6	11/16, 68.8	26/98, 26.5	<0.05	<0.01
Strabismus (n/total, %)	9/35, 25.7	6/47, 12.8	10/16, 62.5	25/98, 25.5	<0.05	<0.01
Gastrointestinal or sensitive disturbances (n/total, %)	4/35, 11.4	4/47, 8.5	10/16, 62.5	18/98, 18.4	>0.05	<0.01
Circadian rhythms and sleep (n/total, %)	NA	3/47, 6.4	8/16, 50.0	11/63, 17.5	-	<0.01
Abnormal pain threshold (n/total, %)	NA	5/47, 10.6	4/16, 25.0	9/63, 14.3	-	<0.01
Cold hands and feet (n/total, %)	6/35, 17.1	2/47, 4.3	7/16, 43.8	15/98, 15.3	>0.05	<0.01
Pacemaker implantation (n/total, %)	3/35, 8.6	1/47, 2.1	0/16, 0	4/98, 4.1	>0.05	>0.05
Abnormal behavior (n/total, %)	NA	17/57, 29.8	14/16, 87.5	31/73, 42.3	-	<0.01
Tumors of neural crest origin (n/total, %)	24/43, 55.8	19/57, 33.3	9/16, 56.3	52/116, 44.8	>0.05	>0.05

*: Group 1 vs Group 3, #: Group 2 vs Group 3