

**Table e-1. Published cases with co-existent Parkinson's disease and 22q11.2 deletion syndrome**

Reference	Year	Cases <sup>a</sup> n = 35	(M/F) (28/7)	Publication	Method of identification
Dufournet, et al. <sup>a</sup>	2017	9	(8/1)	Article	Personal communication
Butcher, et al. <sup>a</sup>	2017	2	(1/1)	Article	Publication by our own group
Lopez-Rivera, et al.	2017	1	(1/0)	Article	Personal communication
Mills and Reich	2016	1	(1/0)	Abstract (poster)	Personal communication
Foo, et al.	2016	1	(0/1)	Article	PubMed
Mok, et al.	2016	8	(6/2)	Article	PubMed/Web of Science
Pollard, et al.	2016	1	(0/1)	Article	PubMed
Oki, et al.	2016	1	(1/0)	Article	PubMed/Web of Science
Perandones, et al. <sup>b</sup>	2015	1	(1/0)	Article	Personal communication
Merico, et al. <sup>a</sup>	2015	3	(2/1)	Article	Personal archive
Rehman, et al.	2015	1	(1/0)	Article	PubMed/Web of Science
Dufournet, et al. <sup>a,c</sup>	2015	9	(8/1)	Abstract (oral)	Web of Science
Verhoeven and Egger	2015	1	(1/0)	Article	Personal archive
Butcher, et al. <sup>a</sup>	2015	1	(1/0)	Abstract (poster)	Web of Science
Boot, et al. <sup>a</sup>	2015	1	(1/0)	Article	PubMed/Web of Science
Butcher, et al. <sup>a</sup>	2014	1	(1/0)	Abstract (poster)	Web of Science
Cheung, et al. <sup>a</sup>	2014	1	(1/0)	Article	Personal archive
Butcher, et al. <sup>a</sup>	2013	4	(3/1)	Article	PubMed/Web of Science
Booij, et al.	2010	1	(1/0)	Article	PubMed
Baudoin, et al. <sup>a</sup>	2010	1	(1/0)	Abstract (poster)	Web of Science
Zaleski, et al. <sup>a</sup>	2009	2	(2/0)	Article	PubMed
Vogels, et al.	2002	1	(1/0)	Article	Personal archive
Arnold, et al. <sup>a</sup>	2001	1	(1/0)	Article	Personal archive
Krahn, et al. <sup>d</sup>	1998	1	(1/0)	Article	PubMed/Web of Science

<sup>a</sup>Thirteen cases were reported two or more times in different publications; <sup>b</sup>Case report of a patient with mosaic 22q11.2 deletion syndrome; <sup>c</sup>10 patients with Parkinson's disease reported; the diagnosis was revoked in one female patient with normal presynaptic dopaminergic imaging; <sup>d</sup>Case report of a patient with a clinically suspected Parkinson's disease diagnosis by a neurologist.

**Table e-2. Lifetime clinical features associated with the 22q11.2 deletion in 45 patients with Parkinson's disease**

		<b>n</b>	<b>%</b>
<b>Intellectual functioning</b>	Normal to borderline	7	15.6
	Mild ID	21	46.7
	Moderate ID	5	11.1
	Severe to profound ID	2	4.4
	Unknown	10	22.2
<b>Schizophrenia spectrum disorder</b>		11	24.4
<b>Seizures/epilepsy</b>	Total	15	33.3
	Symptomatic seizures	7	15.6
	Epilepsy	4	8.9
	Not further classified	4	8.9
<b>Endocrine/metabolic related disease</b>		23	51.1
	Hypocalcemia	16	35.6
	Hypoparathyroidism	5	11.1
	Hypothyroidism	7	15.6
	Diabetes	4	8.9
<b>Congenital heart defect</b>		11	24.4

ID = intellectual disability

**Table e-3. Antiparkinsonian treatment in 35 patients with 22q11.2 deletion syndrome-associated Parkinson's disease at time of last follow-up**

<b>Number of antiparkinsonian agents per patient</b>	<b>n</b>	<b>%</b>
0	3	8.6
1	18	51.4
2	5	14.3
3	7	20.0
4	1	2.9
5	1	2.9
<b>Medications</b>	<b>n</b>	<b>%</b>
Levodopa	29	82.9
Dopamine agonist	11	31.4
Anticholinergic	6	17.1
MAO inhibitor	5	14.3
COMT inhibitor	4	11.4
Amantadine	2	5.7
Selective adenosine <sub>2A</sub> receptor antagonist	1	2.9

Data on medication use at time of last follow-up was available for 35 cases.  
MAO = monoamine oxidase; COMT = catechol-*O*-methyl transferase.