

## SUPPLEMENTARY MATERIAL

Table. Papers and cases included in the study.

Author	Cases reported	Cases included	uncontrolled	controlled	asymptomatic	No outcome or treatment	w/o genotype or phenotype	duplicated cases	ref	PMID
Zhou Q, et al (2014)	9	5	2	3		4			[1]	24552284
Navon Elkan P, et al (2014)	24	21	5	14	2		3		[2]	24552285
Garg N, et al (2014)	1	1	1						[3]	24737293
van Montfrans J, et al (2014)	2	2	1	1					[4]	25075845
Van Eyck L, et al (2014)	2	2		2					[5]	25075846
Bras J, et al (2014)	3	0				3			[6]	25075847
Van Eyck L, et al (2014)	1	1		1					[7]	25075848
Belot A, et al. (2014)	2	2	2						[8]	25278816
Van Eyck L, Jr., et al. (2015)	2	0						2 cases in 25075846	[9]	25457153
Westendorp WF, et al. (2015)	2	1		1		1			[10]	25888558
Gonzalez Santiago TM, et al (2015)	2	2	1		1				[11]	26131734
Batu ED, et al (2015)	6	3	1	2				3 cases in 24552284 (16-18)	[12]	26233953
Fellmann F, et al. (2016)	2	2	2						[13]	26607704

Van Montfrans JM, et al (2016)	9	5		5			4 cases of 25888558	[14 ]	26867732
Poswar Fde O, et al (2016)	2	2	1		1			[15 ]	26914925
Schepp J, et al (2016)	2	2	2					[16 ]	26922074
Nanthapisal S, et al (2016)	15	15		10	5			[17 ]	27059682
Keer N, et al (2016)	1	1		1				[18 ]	27069017
Hsu AP, et al. (2016)	1	1		1				[19 ]	27130863
Uettwiller F, et al. (2016)	2	2		2				[20 ]	27252897
Pichard DC, et al. (2016)	1	0					1 case in 24552284	[21 ]	27444081
Ben-Ami T, et al. (2016)	5	4	4			1		[22 ]	27514238
Elbracht M, et al. (2017)	1	1		1				[23 ]	28024309
Hashem H, et al. (2017)	1	0					One case in 28974505	[24 ]	28230570
Schepp J, et al. (2017)	11	5	3	2		4	2 cases in 26922074	[25 ]	28493328
Sahin S, et al (2018)	8	5	2	3			One case in 24552285, one in 24737293 and the 3 <sup>rd</sup> case in 30642227	[26 ]	28516235
Caorsi R, et al. (2017)	17	17		16	1			[27 ]	28522451
Nihira H, et al (2018)	1	1		1				[28 ]	28665179
Hashem H, et al. (2017)	1	0					One case in 28974505	[29 ]	28805790

Skrabl-Baumgartner A, et al (2017)	2	2	2					[30]	28830446
Hashem H, et al. (2017)	14	10		10			One in 27130863, 2 <sup>nd</sup> in 26867732, 3 <sup>rd</sup> in 25075846 and 4 <sup>th</sup> in 25075845	[31]	28974505
Buccioli G, et al. (2017)	1	0					1 case in 25075846	[32]	28993957
Alsultan A, et al (2018)	1	1		1				[33]	29271561
Trotta L, et al. (2018)	9	4	2	1	1	5		[34]	29391253
Gunthner R, et al. (2018)	1	1		1				[35]	29391272
Michniacki TF, et al. (2018)	2	2		1	1			[36]	29411230
Cipe FE, et al. (2018)	1	1	1					[37]	29564582
Lamprecht P, et al (2018)	1	1		1				[38]	29600946
Sundin et al (2019)	1	1		1				[39]	29620681
Rama M et al (2018)	13	0				12	One case in in 28522451 (case 5)	[40]	29681619
Teixeira V, et al (2018)	1	1		1				[41]	29916967
Springer JM, et al. (2018)	2	2		2				[42]	29963054
Van Nieuwenhove E, et al. (2018)	2	1			1		One case in 25075848	[43]	30139808
Maccora I, et al (2018)	1	1		1				[44]	30148442

Sharma A, et al. (2019)	1	1		1			[45 ]	30165497
Kisla Ekinici RM, et al. (2018)	1	1		1			[46 ]	30377239
Arts K, et al. (2018)	3	3	2		1		[47 ]	30386947
Martin H, et al (2018)	2	2	1	1			[48 ]	30514670
Claassen D, et al. (2018)	1	1	1				[49 ]	30559313
Liu L, et al (2019)	1	1		1			[50 ]	30610243
Sahin et al (2019)	2	1		1		1	[51 ]	30642227
Alabbas F, et al. (2019)	2	2	1	1			[52 ]	30644014
Insalaco A, et al. (2019)	5	1		1		4 cases in 28522451	[53 ]	30647181
Barzaghi F, et al (2018)	1	1		1			[54 ]	30692987
Grossi A, et al. (2019)	1	1		1			[55 ]	30920658
Ghurye RR, et al (2019)	2	2	1	1			[56 ]	30924144
Goncalves TDS, et al. (2019)	1	0			1		[57 ]	30962308
Gibson KM, et al (2019)	11	8	3	5		3	[58 ]	31008556
Ozen S, et al (2020)	24	18	5	13		6 cases in 26233953	[59 ]	31043544
Liebowitz J,et al. (2019)	3	3		3			[60 ]	31092714
Clarke K, et al. (2019)	1	1		1			[61 ]	31291964

Kisla Ekinci RM, et al. (2020)	5	4		4			one case in 30377239	[62 ]	31292637
Ekinci RMK et al (2019)	1	1		1				[63 ]	31522599
Sozeri B, et al. (2019)	5	4		4			Once case in 28516235	[64 ]	31541281
Akgun-Dogan O, et al. (2019)	1	1	1					[65 ]	31584751
Alaygut D, et al. (2019)	1	1	1					[66 ]	31598601
Chong-Neto HJ, et al. (2019)	1	1		1				[67 ]	31617030
Krutzke S et al. (2019)	2	2		2				[68 ]	31651641
Goschl L, et al. (2020)	1	1		1				[69 ]	31686313
Tanatar A, et al. (2020)	5	4		4			One case in 31916720	[70 ]	31848804
van Well GTJ, et al. (2019)	6	6	1	5				[71 ]	31856934
Cakan M, et al. (2019)	1	1		1				[72 ]	31916720
Lee PY, et al. (2020)	15	15	14	1				[73 ]	31945408
Ganhão S, et al (2020)	2	2	1	1				[74 ]	32535845
Schnappauf O, et al (2020)	6	6	1	3	2			[75 ]	32638197
Saettini F, et al (2020)	2	1			1	1		[76 ]	32659374
Sarrabay G, et al. (2015)	5	0					5 cases in 29681619	[77 ]	L609058053
Benner MF, et al. (2016)	1	0					case in 26867732	[78 ]	L619272204

Chang Y, et al. (2018)	1	1		1					[79 ]	L622389226
Li J, et al. (2018)	1	1	1						[80 ]	L624305135
Schnider C, et al. (2018)	1	1		1					[81 ]	L625167642
Naidu GSRSNK, et al. (2018)	3	1		1		1		One case in 30165497	[82 ]	L626163913
Garbarino F, et al. (2019)	1	1		1					[83 ]	L627973706
Kilic SS, et al. (2019)	1	0						same case in L629479534	[84 ]	L628681361
Paola K, et al. (2019)	2	2		2					[85 ]	L628807871
Ouail DE, et al. (2019)	3	0			1	2			[86 ]	L628809641
Kilic SS, et al. (2019)	1	1	1						[87 ]	L629479534
Al Mosawi Z, et al. (2019)	1	1		1					[88 ]	L629806493
Caorsi R, et al. (2014)	3	0						3 cases in 28522451	[89 ]	L71686775
Van Montfrans J, et al. (2014)	2	0						2 cases in 25075845	[90 ]	L71778687
Toz B, et al. (2015)	1	1		1					[91 ]	L72064769
Sasa GS, et al. (2015)	2	0				1		1 case in 28974505	[92 ]	L72174305
Loureiro G, et al. (2019)	1	0						1 case in 32535845	[93 ]	L628841823
Neishabury M (2020)	2	2	1	1					[94 ]	31097629
Our cases	2	2		2						
<b>Total</b>	<b>338</b>	<b>242</b>	<b>68</b>	<b>157</b>	<b>17</b>	<b>34</b>	<b>10</b>	<b>52</b>		

Note: A total of 94 papers were enclosed, reporting 336 cases. 52 cases were excluded due to repeated reporting in different papers and 10 cases were excluded due to unconfirmed genotype or lack of detailed phenotype information. A further 34 cases were excluded due to lack of outcome or treatment report. Along with 2 cases from our center, a total of 242 cases were enclosed, with 17 patients were asymptomatic requiring no treatment, 68 patients had uncontrolled disease after treatment and 157 patients reached disease control after treatment.

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