Article title: Evaluation of the X-chromosome inactivation patterns in females with Gabriele-de Vries syndrome and expansion of clinical spectrum

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Table S1 Summary of published cases of Gabriele-de Vries syndrome (GDVS). The table includes publication of reference, number of novel cases reported, identification of individual patients when more than one case is reported, variant nomenclature, variant type and reported classification of pathogenicity, inheritance pattern, the presence or absence of a clinical description, age of referral or examination and sex of the affected individuals, the presence or absence of neurodevelopmental disorders, movement abnormalities, hormone/thyroid dysfunction and/or autoimmune conditions, and additional observations

Publication	Number of novel reported cases of GDVS	Identification (if applicable) and variant	Variant type	Reported classification	Segregation analysis	Clinical description available	Age at referral or examination	Sex	Neurodevelopmental disorders (summary)	Movement abnormalities (summary)	Hormone/thyroid dysfunction and/or autoimmune conditions (summary)	Additional observations
Gabriele et al., 2017	10	**Individual 1** c.1138G>T p.Asp380Tyr	missense	pathogenic	*de novo*	+	2 years, 9 months	male	+ (global development delay, moderate speech delay, delayed motor development)	-	NA	some neurodevelopmental features cannot be evaluated due to the age of the patient
		Individual 2 c.1097T>C p.Leu366Pro	missense	pathogenic	*de novo*	+	15 years, 10 months	male	+ (moderate intellectual disability, first words at 2 years, delayed motor development)	+ (toe walking)	NA	
		Individual 3 c.1096C>G p.Leu366Val	missense	pathogenic	*de novo*	+	5 years, 1 month	female	+ (mild intellectual disability, mildly delayed speech development, delayed motor development)	-	NA	
		Individual 4 c.1030C>T p.Gln344*	nonsense	pathogenic	*de novo*	+	39 years	female	+ (mild intellectual disability, mildly delayed speech development, delayed motor development)	+ (tremor)	NA	

Table S1 (continued)

Publication	Number of novel reported cases of GDVS	Identification (if applicable) and variant	Variant type	Reported classification	Segregation analysis	Clinical description available	Age at referral or examination	Sex	Neurodevelopmental disorders (summary)	Movement abnormalities (summary)	Hormone/thyroid dysfunction and/or autoimmune conditions (summary)	Additional observations
Gabriele et al., 2017 (continued)		**Individual 5** c.535A>T p.Lys179*	nonsense	pathogenic	*de novo*	+	17 years, 6 months	female	+ (mild intellectual disability, learning difficulties, moderate speech delay, delayed motor development)	+ (progressive dystonia)	+ (hypothyroidism)	
		Individual 6 c.1173delT p.Asn391Lysfs*10	frameshift	pathogenic	*de novo*	+	7 years, 10 months	male	+ (mild to moderate intellectual disability, speech delay, mildly delayed motor development)	+ (waddling gait)	NA	
		Individual 7 c.1174_1176del p.Lys393del	missense	pathogenic	*de novo*	+	1 year, 3 months	male	+ (mild global development delay	-	NA	some neurodevelopmental features cannot be evaluated due to the age of the patient
		Individual 8 c.385delG p.Asp129Ilefs*127	frameshift	pathogenic	*de novo*	+	35 years	female	+ (mild global developmental delay, delayed speech development, delayed motor development)	+ (progressive dystonia, progressive gait impairment, dysarthria)	+ (hypothyroidism)	individual 8 was also later described by Ferng et al. (2022)
		Individual 9 c.1015A>C p.Lys339Gln	missense	pathogenic	*de novo*	+	9 years, 3 months	male	+ (moderate to severe intellectual disability, non-verbal, delayed motor development)	-	+ (growth hormone deficiency)	

Table S1 (continued)

Publication	Number of novel reported cases of GDVS	Identification (if applicable) and variant	Variant type	Reported classification	Segregation analysis	Clinical description available	Age at referral or examination	Sex	Neurodevelopmental disorders (summary)	Movement abnormalities (summary)	Hormone/thyroid dysfunction and/or autoimmune conditions (summary)	Additional observations
Gabriele et al., 2017 (continued)		**Individual 10** c.958C>T p.His320Tyr	missense	pathogenic	*de novo*	+	1 year, 5 months	female	+ (delayed motor development)	-	NA	some neurodevelopmental features cannot be evaluated due to the age of the patient
Morales-Rosado et al., 2018	1	c.860_864delTTAAAA p.Ile287Arg fs*3	frameshift	presumed pathogenic	*de novo*	+	25 years	female	+ (delayed motor development, learning disability)	+ (wide-based, waddling, somewhat antalgic gait)	+ (autoimmune myasthenia gravis)	
Carminho-Rodrigues et al., 2020	1	(NM: 003403.4) c.907T>C p.Cys303Arg	missense	pathogenic	*de novo*	+	21 years	female	+ (mild intellectual deficit)	+ (complex movement disorder including an action tremor, cerebellar ataxia, dystonia, and partial ocular apraxia as the most striking feature)	-	this patient was included in the present study (P9)
Bae et al., 2021	1	c.1220A>G p.His407Arg	missense	likely pathogenic	*de novo*	+	7 months	female	+ (mild developmental delay)	NA	-	some motor and neurodevelopmental features cannot be evaluated due to the age of the patient
Balakrishnan & Ranganath, 2021	1	(NM_003403) c.690del p.Asp231IlefsTer25	frameshift	pathogenic	NA	+	11 years	female	+ (moderate intellectual disability, global developmental delay)	+ (altered gait with recurrent falls, owing to right-sided curvature of the spine and associated dystonia, twisting movements of lower limbs and hip joints, abnormal posturing of hands)	NA	

Table S1 (continued)

Publication	Number of novel reported cases of GDVS	Identification (if applicable) and variant	Variant type	Reported classification	Segregation analysis	Clinical description available	Age at referral or examination	Sex	Neurodevelopmental disorders (summary)	Movement abnormalities (summary)	Hormone/thyroid dysfunction and/or autoimmune conditions (summary)	Additional observations
Zorzi et al., 2021	1	(NM_003403) c.1118_1119delAT p.His373Argfs*18	frameshift	pathogenic	*de novo*	+	32 years	male	+ (moderate intellectual disability, slight delay of psychomotor development, learning difficulties)	+ (severe generalized dystonia with continuous dystonic movements, prominent trunk involvement, and abduction-type laryngeal dystonia)	NA	
Tan et al., 2021	1	(NM_003403.5) c.1124G>A p.Arg375Gln	missense	likely pathogenic	*de novo*	+	9 months	female	+ (developmental delay)	NA	-	some motor and neurodevelopmental features cannot be evaluated due to the age of the patient
Malaquias et al., 2021	1	(NM_003403.4) c.1099dup p.Asp367Glyfs*25	frameshift	pathogenic	*de novo*	+	38 years	female	-	+ (writer's clamp, generalized dystonia, with severe oromandibular involvement)	-	
Imafidon et al., 2021	1	**ID 659** c.568_581del p.Ala190Argfs*34	frameshift	pathogenic	*de novo*	+ (not much detailed)	NA	male	NA	NA	NA	it is a large study that does not provide a detailed description of each case
Zech et al., 2021	1	**Patient 39** (NM_003403.4) c.1118A>G p.His373Arg	missense	NA	NA	-	NA	NA	NA	+ (dystonia)	NA	it is a large study of a cohort of individuals with dystonia, with no detailed general description of each case

Table S1 (continued)

Publication	Number of novel reported cases of GDVS	Identification (if applicable) and variant	Variant type	Reported classification	Segregation analysis	Clinical description available	Age at referral or examination	Sex	Neurodevelopmental disorders (summary)	Movement abnormalities (summary)	Hormone/thyroid dysfunction and/or autoimmune conditions (summary)	Additional observations
Ferng et al., 2022	1	**Case 1** c.908G>T p.Cys303Phe	missense	NA	NA	+	22 years	female	+ (mild global developmental delay, autism)	+ (generalized dystonia involving feet, hands, torso, tongue, jaw, pharynx, and larynx, worsening dysarthria, gait abnormality)	NA	the other case described in the article (Case 2) had been previously published by Gabriele et al. (2017)
Khamirani et al., 2022	1	(NM_003403) c.690delA p.Glu231Ilefs*25	frameshift	pathogenic	*de novo*	+	10 years	male	+ (developmental delay, motor delay, moderate hypotonia, cerebral atrophy, severe learning disability, autistic behavior, ADHD)	+ (gait abnormality, ataxia, ocular movement disorder)	NA	
Cherik et al., 2022	12	**YY1-1** (NM_003403.4) c.1007A>G p.Glu336Gly	missense	NA	*de novo*	+	18 years, 2 months	female	+ (motor developmental delay, intellectual disability)	-	-	
		YY1-2 (NM_003403.3) c.1112G>A p.Arg371His	missense	NA	*de novo*	+	16 years, 7 months	female	+ (motor developmental delay, intellectual disability)	-	-	
		YY1-3 YY1arr[GRCh37]14q32.2(100402364-101351127)x1	CNV deletion	NA	*de novo*	+	31 years, 9 months	male	+ (motor developmental delay, intellectual disability)	+ (severe dystonia, spasticity)	-	

Table S1 (continued)

Publication	Number of novel reported cases of GDVS	Identification (if applicable) and variant	Variant type	Reported classification	Segregation analysis	Clinical description available	Age at referral or examination	Sex	Neurodevelopmental disorders (summary)	Movement abnormalities (summary)	Hormone/thyroid dysfunction and/or autoimmune conditions (summary)	Additional observations
Cherik et al., 2022 (continued)		**YY1-4** (NM_003403.4) c.1001T>C p.Phe334Ser	missense	NA	*de novo*	+	6 years	female	+ (motor developmental delay, intellectual disability)	+ (dystonia)	-	
		YY1-5 (NM_003403.4) c.1151_1154dup p.Pro386Valfs*7	frameshift	NA	*de novo*	+	6 years, 2 months	female	+ (motor developmental delay, intellectual disability)	-	NA	
		YY1-7 (NM_003403.5) c.1067C>T p.Thr356Met	missense	NA	paternal	+	5 years, 9 months	male	+ (motor developmental delay, intellectual disability)	-	-	
		YY1-8 c.1121T>G p.Val374Gly	missense	NA	*de novo*	+	4 years, 9 months	male	+ (motor developmental delay)	-	-	some neurodevelopmental features cannot be evaluated due to the age of the patient
		YY1-9 YY1:c.959A>G p.His320Arg	missense	NA	??	+	2 years, 1 months	male	+ (motor developmental delay, global developmental delay)	-	NA	some neurodevelopmental features cannot be evaluated due to the age of the patient
		YY1-11 (NM_003403.5) c.690dup p.Asn231Argfs*3	frameshift	NA	*de novo*	+	8 years, 1 months	male	+ (motor developmental delay, learning difficulties, facial tics, Tourette syndrome)	+ (gait imbalance, exercise-induced muscle fatigue)	-	

Table S1 (continued)

Publication	Number of novel reported cases of GDVS	Identification (if applicable) and variant	Variant type	Reported classification	Segregation analysis	Clinical description available	Age at referral or examination	Sex	Neurodevelopmental disorders (summary)	Movement abnormalities (summary)	Hormone/thyroid dysfunction and/or autoimmune conditions (summary)	Additional observations
Cherik et al., 2022 (continued)		**YY1-12** (NM_003403.5) c.1124G>A p.Arg375Gln	missense	NA	*de novo*	+	25 years, 7 months	male	+ (motor developmental delay, intellectual disability)	-	+ (hypothyroidism)	
		YY1-13 (NM_003403.4) c.908G>T p.Cys303Phe	missense	NA	*de novo*	+	20 years, 6 months	male	+ (motor developmental delay, intellectual disability)	+ (generalized dystonia)	+ (thyroid nodule)	
		YY1-6 (NM_003403.5) c.1067C>T p.Thr356Met	missense	NA	*de novo*	-	33 years	male	+ (intellectual disability)	-	-	
dos Santos et al., 2022	1	(NM_003403.5) c.1106A>G p.Asn369Ser	missense	pathogenic	*de novo*	+	22 years	female	+ (global developmental delay, intellectual disability)	-	-	this patient was included in the present study (P10)
Alali & Vitalone, 2022	1	c.769G>T p.Gly257*	nonsense	pathogenic	*de novo*	+	10 years	female	+ (global developmental delay, ADHD)	-	+ (autoimmune hypothyroidism)	
Chaves et al., 2023	1	(NM_003403.5) c.1062+1G>A NP_003394.1:p.?	splicing	pathogenic	*de novo*	+ (not much detailed)	8 years, 11 months	female	+ (intellectual disability, ADHD, learning disability)	-	+ (atopic dermatitis, Hashimoto's thyroiditis)	this patient was included in the present study (P11); atopic dermatitis and Hashimoto's thyroiditis were identified upon re-evaluation (not reported in Chaves et al., 2023)

Table S1 (continued)

Publication	Number of novel reported cases of GDVS	Identification (if applicable) and variant	Variant type	Reported classification	Segregation analysis	Clinical description available	Age at referral or examination	Sex	Neurodevelopmental disorders (summary)	Movement abnormalities (summary)	Hormone/thyroid dysfunction and/or autoimmune conditions (summary)	Additional observations
Koruga et al., 2023	1	c.1A>C p.Met1?	start loss	likely pathogenic	*de novo*	+	NA	male	+ (mildly delayed motor and speech development, slow psychomotor development)	-	NA	
Woo; Kim; Kim, 2023	1	c.1130A>G p.His377Arg	missense	pathogenic	*de novo*	+	4 years	female	+ (global developmental delay, cognitive impairment, autistic features)	-	NA	some neurodevelopmental features cannot be evaluated due to the age of the patient
Chawla et al., 2023	1	c.625C>T p.Gln209Ter	nonsense	likely pathogenic	NA	+	12 years	female	-	+ (writer's cramp and myoclonic jerks at 9 years, evolving to action induced dystonia in the right upper limb, followed by similar movements in the left upper limb as well)	NA	
Asato et al., 2023	1	c.1043C>T p.Thr348Ile	missense	likely pathogenic	*de novo*	+	8 months	male	+ (gross motor delay)	NA	+ (hypothyroidism)	some motor and neurodevelopmental features cannot be evaluated due to the age of the patient
Yang et al., 2024	1	(NM_003403.5) c.458_476del p.Val153Alafs*97	frameshift	pathogenic	NA	+	1 year, 10 months	female	+ (motor developmental delay, delayed language development)	-	-	some neurodevelopmental features cannot be evaluated due to the age of the patient

Table S1 (continued)

Publication	Number of novel reported cases of GDVS	Identification (if applicable) and variant	Variant type	Reported classification	Segregation analysis	Clinical description available	Age at referral or examination	Sex	Neurodevelopmental disorders (summary)	Movement abnormalities (summary)	Hormone/thyroid dysfunction and/or autoimmune conditions (summary)	Additional observations
Srour et al., 2024	1	(NM_003403.4) c.1024C>T p.Arg342*	stop gain	VUS	NA	+	1 year, 4 months	male	+ (delayed developmental milestones, speech delay)	+ (lack of coordination, ataxic gait)	NA	some neurodevelopmental features cannot be evaluated due to the age of the patient
Shin et al., 2024	1	(NM_003403.5) c.843-1_863del	splice site	likely pathogenic	NA	+	28 years	male	-	+ (limb dystonia, gait disturbance, infantile nystagmus, tremors)	-	
Topa et al., 2024	1	**P2605_115(M)** c.1057T>C p.Phe353Leu	missense	likely pathogenic	expected *de novo* (absent in the mother, father unavailable)	-	NA	male	NA	NA	NA	it is a large study of a cohort of individuals with craniosynostosis, with no detailed general description of each case
Luo et al., 2024	1	**NG21-81** (NM_003403.4) c.682delG p.Glu228LysfsTer 28	frameshift	likely pathogenic	*de novo*	-	gestational week: 24	NA	NA	NA	NA	it is a large study of a cohort of foetuses with central nervous system abnormalities
Mudassir et al., 2025	1	**ES-10** (NM_003403.5) c.141_143delGGA p.Glu47del	in-frame deletion	pathogenic	*de novo*	+	7 years, 8 months	male	+ (IQ < 25)	+ (movement disorder including ataxia and seizures)	NA	
Pal et al., 2025	1	(NM_003403.5) c.1025G>A p.Arg342Gln	missense	pathogenic	*de novo*	+	21 years	female	+ (global developmental delay)	-	+ (autoimmune hypothyroidism)	

Table S1 (continued)

Publication	Number of novel reported cases of GDVS	Identification (if applicable) and variant	Variant type	Reported classification	Segregation analysis	Clinical description available	Age at referral or examination	Sex	Neurodevelopmental disorders (summary)	Movement abnormalities (summary)	Hormone/thyroid dysfunction and/or autoimmune conditions (summary)	Additional observations
Huang et al., 2025	1	c.385del p.Asp129Ilefs*127	frameshift	pathogenic	*de novo*	+	9 years, 5 months	female	+ (mild intellectual disability)	-	+ (Hashimoto's thyroiditis)	

Abbreviations: +: present; -: absent; ADHD: Attention Deficit Hyperactivity Disorder; IQ: intelligence quotient; NA: information not available

Table S2 Summary of the clinical traits of the eleven patients with heterozygous pathogenic/likely pathogenic variants in *YY1* (three of them previously reported) who were evaluated in this study

	Patient 1	**Patient 2**	**Patient 3**	**Patient 4 (mother of Patient 5)**	**Patient 5**	**Patient 6**	**Patient 7**	**Patient 8**	**Patient 9**	**Patient 10**	**Patient 11**
Global developmental delay	+	+	+	+	+ Neonatal axial hypotonia (premature birth at 30+3 WG)	+	Delayed motor and socio-emotional development, speech development is not delayed	+	+	+	-
Intellectual disability	not applicable (< 5 years old)	not applicable (< 5 years old)	+ (mild)	+ (mild)	NA (too young)	+	+ (IQ 82)	+	+	+ (moderate to severe)	+ (mild)
Delayed speech and language development	+	+	+	+	+	+	-	NA (parents do not recall)	+	+	+
Attention Deficit Hyperactivity Disorder (ADHD)	+	-	+	-	NA (too young)	+	+ (Attention Deficit Disorder)	NA	+	NA	+
Autism Spectrum Disorder (ASD)	+ (nonverbal)	under investigation	-	-	NA (too young)	-	-	NA	-	+	+ (diagnosed at 18 years of age)
Anxiety	NA	-	+	-	NA (too young)	-	-	NA	+	+	-
Self-injurious behavior	+	-	-	-	NA (too young)	-	-	NA	-	+	+ (trichotillomania)
Sleep disturbance	+	+	-	-	-	-	-	NA	-	-	+ insomnia (screen)
Ataxia/unsteady gait	+	+	+	- (dizziness)	NA (too young)	+	-	NA	+	wheelchair	-
***Corpus callosum* malformations**	+ (thin *corpus callosum*)	-	NA	-	-	-	NA MRI not performed	NA	-	-	-
Hydrocephalus	NA	+ (modest)	NA	-	-	-	-	NA	-	-	-
Impairment of visual pursuit	+	NA	-	-	- (retinopathy due to prematurity)	-	-	NA	+	-	-
Strabismus	+ (convergent)	-	-	+ (left eye)	+	-	-	-	+	+	+ (surgery at the age of 10 years)
Hearing impairment	+ (bilateral neurosensory)	NA	-	-	-	-	-	NA	-	+ (mild)	-
Dysarthria	- (no speech)	+	NA	-	NA (too young)	+	-	NA	+	- (no speech)	-
Thyroid dysfunction	-	-	+ (Hashimoto's thyroiditis)	-	-	-	+ (Graves' Disease)	+ (hyperthyroidism)	-	-	+ (Hashimoto's thyroiditis)

Table S2 (continued)

	Patient 1	Patient 2	Patient 3	Patient 4 (mother of Patient 5)	Patient 5	Patient 6 (France)	Patient 7	Patient 8	Patient 9	Patient 10	Patient 11
Feeding difficulties	+	-	-	-	-	+	+ (dystrophy due to severe feeding difficulties with swallowing difficulties and recurrent vomiting, inefficient motility of the esophagus)	+	-	+	-
Gastroesophageal reflux	+	-	-	-	-	+	+	+	-	+ (as a baby) (abdominal migraine and vomiting)	-
Constipation	+	-	-	-	-	+	-	+	-	+	+
Recurrent infections	+	-	+	-	NA (too young)	-	+	+ (Upper Respiratory Infections and acute gastroenteritis)	-	+	sinus infection (3-8 years)
Craniofacial asymmetry	+	NA	NA	-	- (brachycephaly)	+	+	+	+	- (microcephaly long face)	-
Large/prominent forehead	+	+	+	+	+	+	-	+	+	+	-
Malar flattening	+	NA	NA	-	-	+	+	+	+	+	-
Abnormal ear morphology	+ (low-set, posteriorly rotated ears)	NA	+ (simple ears and overfolded helix)	-	+ (low set ears, overfolded helix)	+ (low-set, posteriorly rotated ears)	+ (protruding ears)	-	+	+	+ (R preauricular pit)
Periorbital fullness	+	+	+	+	+	-	+	+	+	+	+
Downslanted palpebral fissures	+	+	+	-	-	+	-	+	+	+	-
Eyelid ptosis	+	+	+ (bilateral)	-	-	-	-	-	-	+	-
Proptosis	+	-	+	-	-	+	-	+	-	-	-
Epicanthus	-	NA	+	-	-	-	-	NA	-	+	+
Telecanthus	+	NA	+	-	-	+	-	NA	-	+	hypertelorism
Almond-shaped eyes	+	NA	NA	+	+	+	+	NA	+	-	+
Wide, concave nasal bridge	+	NA	+	-	-	+	-	NA	+	+	-
Bulbous nose tip	+	NA	+	+	+	+	-	+	+	-	-
Abnormal columella	+ (short)	NA	NA	+ (enlarged columella)	+ (enlarged columella)	+ (short)	+ (short)	NA	+ (short)	+ (short)	-

Table S2 (continued)

	Patient 1	Patient 2	Patient 3	Patient 4 (mother of Patient 5)	Patient 5	Patient 6 (France)	Patient 7	Patient 8	Patient 9	Patient 10	Patient 11
Downturned corners of mouth	+	+	NA	-	-	+	-	+	+	-	-
Thick lower lip	-	-	+	-	-	-	-	+ (thick lips)	+	-	+
Micrognathia	+	+	+	-	-	+	-	NA	-	-	+
Pointed chin	+	+	+	+	+	+	-	-	+	+	-
Sparse hair	+	+	NA	-	+	+	-	-	-	-	-
Arachnodactyly	+	NA	+	-	-	-	-	+	-	+	+
Camptodactyly	-	NA	+	-	-	-	-	NA	-	-	-
Fingers with joints hypermobility	+	NA	+	-	-	+	-	NA	-	+	-
Tapered phalanx of fingers	NA (radiography results not available)	NA	+	-	-	-	-	NA	-	-	NA
Marfanoid habitus	+	NA	+	-	-	+	-	NA	+	+	+
Big hands and feet	+	-	+	-	-	-	-	+	-	+ (hands)	-
Pregnancy complications (other than intrauterine growth restriction)	- (prenatal diagnosis of intrauterine growth restriction)	+ (maternal diabetes)	+ (maternal hypertension)	+ (maternal hypertension)	+ twin pregnancy, preeclampsia	-	-	(anemia)	-	-	-
Intrauterine growth restriction	+	+	+	NA	+ (twin bichorial biamniotic pregnancy)	+	-	NA	-	+	-
Premature birth	+	-	+	+	+	+	-	-	-	-	-
Low birth weight	+	+	+	+	+	+	+ (3 P., -1.91 z)	+	+ (3rd centile)	-	-
Neonatal jaundice	-	-	+ (treated with phototherapy)	NA	+ (treated with phototherapy)	-	-	NA	-	+	-
Congenital torticollis	-	+	-	-	-	-	-	NA	-	-	-
Nasolacrimal duct obstruction	+	NA	+ (treated by surgical intervention)	-	-	-	-	NA	-	-	-
Recurrent Moro reflex	+	+	NA	NA	NA	NA	-	NA	-	-	NA
Poor pincer grasp	+	+	NA	NA	NA	-	-	NA	+	+	NA
Hypotonia	+ (congenital)	+ (2-3 years of age)	+ (congenital)	NA	+	+ (congenital)	-	NA	+	+	+

Table S2 (continued)

	Patient 1	Patient 2	Patient 3	Patient 4 (mother of Patient 5)	Patient 5	Patient 6 (France)	Patient 7	Patient 8	Patient 9	Patient 10	Patient 11
Decreased body weight	+	-	-	-	+ (-4 SD)	+	+ (-3.07 z)	+	+	+	+
Short stature	-	-	+	+	+ (-4 SD)	-	-	-	-	+	-
Atopic dermatitis	+	NA	-	-	NA	-	-	NA	-	-	+
Hypertrichosis	+	NA	NA	-	-	-	-	NA	-	-	-
Sinus tachycardia	-	-	-	-	-	-	-	+	-	-	-
Left anterosuperior fascicular block	-	-	-	-	-	-	-	+	-	-	-
Tricuspid regurgitation	-	-	-	-	-	-	-	+	-	-	-
Other				obesity	bilateral single palmar crease, smooth and prominent philtrum, thin upper lip, faded cupid's bow		IgA deficiency, orientation difficulties	fingers with verrucous lesions		severe scoliosis and chorioretinitis	scoliosis (10 years)
Reference	newly identified (*de novo*)	newly identified (*de novo*)	newly identified (*de novo*)	newly identified (*de novo*)	newly identified (inherited from affected parent)	newly identified (*de novo*)	newly identified (*de novo*)	newly identified (*de novo*)	Carminho-Rodrigues *et al.*, 2020 (*de novo*)	dos Santos *et al.*, 2022 (*de novo*)	Chaves *et al.*, 2023 (*de novo*)

+: present; -: absent; NA: information not available; WG: weeks of gestation; MRI: magnetic resonance imaging; IQ: intelligence quotient; SD: standard deviation; P: percentile; z: z-score

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