Characterization of Apparently Balanced Chromosomal Rearrangements from the Developmental Genome Anatomy Project

Anne W. Higgins, Fowzan S. Alkuraya, Amy F. Bosco, Kerry K. Brown, Gail A.P. Bruns, Diana J. Donovan, Robert Eisenman, Yanli Fan, Chantal G. Farra, Heather L. Ferguson, James F. Gusella, David J. Harris, Steven R. Herrick, Chantal Kelly, Hyung-Goo Kim, Shotaro Kishikawa, Bruce R. Korf, Shashikant Kulkarni, Eric Lally, Natalia T. Leach, Emma Lemyre, Janine Lewis, Azra H. Ligon, Weining Lu, Richard L. Maas, Marcy E. MacDonald, Steven D.P. Moore, Roxanna E. Peters, Bradley J. Quade, Fabiola Quintero-Rivera, Irfan Saadi, Yiping Shen, Jay Shendure, Robin E. Williamson, and Cynthia C. Morton*

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In the version of this paper originally published online March 6, 2008, the karyotypes listed in column 3 (heading: Karyotype) for the following cases contained errors: DGAP006, 012, 090, 100, 112, 123, 127, 128, 137, 157, 159, 166, 169, and 174. The corrected table appears below. The authors regret the errors.

Table 1. Phenotype and Karyotype of Selected DGAP Cases

Case No.	Phenotype	Karyotype	Publication
DGAP003	Delayed dentition, gingival hyperplasia, hirsutism, large facial bones and mandibles, large ears, a markedly enlarged nose with short columella nasi and saddle deformity, depressed nasal bridge, hypertelorism with bilateral convergent strabismus, epicanthal folds, protruding upper lip, hypertrophic papillae on the posterior of the tongue, bilateral spade-like fingers, skin thickening on the legs, dysmorphic skeletal features	46,XY,t(3;17)(p14.3;q24.3)dn	21, 33
DGAP006	Mental retardation, developmental delay, absent speech, aggressive behavior, frontal bossing, epicanthal folds, left eye ptosis, low-set ears, no binocular fixation searching movements	46,XX,t(1;2)(p32;q13)dn	
DGAP009	Mental retardation, eye anomaly, other multiple congenital anomalies	46,XY,t(1;8)(p34;q22)dn	
DGAP011	Kallmann syndrome (atrophic testes, azoospermia, cleft lip and palate)	46,XY,t(7;8)(p12.3;p11.2)dn	2
DGAP012	Developmental delay, digitalized thumbs, brachycephaly, microcephaly, small down-turned mouth, mild midfacial hypoplasia, flat mid-face, narrow nasal bridge, very small nose, large ears, bilateral epiblepharon without trichiasis, small hands and feet, absence of emotional expression, hand flapping, early feeding problems	46,XY,t(11;19)(p11.2;p13.3)dn	
DGAP015	Bannayan-Riley-Ruvalcaba syndrome, malignant intracranial hCG-secreting tumor causing precocious puberty	46,XY,t(10;13)(q23.3;q33)dn	
DGAP016	Hypoplastic testes	46,XY,t(8;10)(p11.2;p13)dn	
DGAP018	Bilateral osseocutaneous syndactyly of the 3rd, 4th, and 5th fingers; hypotonia; macrocephaly; forehead and occipital prominence; left inner thigh hemangioma; developmental delay	46,XX,?dup(2)(p14p?21),ins(2;1)(?p13;p21p31),ins(3;2)(q23;p14p?21)dn	
DGAP020	Sex reversal, gonadoblastoma, streak gonad, amenorrhea	46,X,t(Y;17)(q11;p13)dn	
DGAP025	Developmental delay, scoliosis, syndactyly of toes, learning problems, masculinized face, hirsutism	46,X,t(X;15)(p22;q26)dn	
DGAP032	Kallmann syndrome (hypogonadotropic hypogonadism, and anosmia), skeletal anomalies, mental retardation	46,XY,t(7;12)(q21.13;q24)dn	
DGAP089	Subarachnoid hemorrhage, ventriculomegaly, underdeveloped corpus callosum, tonic-clonic seizure, severe delays in growth and development, craniofacial disproportion and dysmorphism, right cryptorchidism, hypotonia, chronic intestinal obstruction	46,XY,t(1;2)(p31.3;q22.1),del(2)(q14.3q21)dn	23
DGAP090	Sensorineural hearing loss, Mondini defect, avascular necrosis of the left femoral head, dermal telangiectasias with ulceration, juvenile rheumatoid arthritis	46,XY,t(8;9)(q13;p22),t(9;11)(q33;q13)mat	34
DGAP095	Seizures, developmental delay, infantile hypotonia, obesity, *livedo reticularis*	46,X,t(X;2)(p11.2;q37)dn	32
DGAP097	Developmental delay, infantile spasms, hypotonia, mental retardation, behavioral problems, facial dysmorphism, myopia, patchy skin hypopigmentation	46,X,t(X;9)(p22.2;p13)dn	

(*Continued on next page*)

Table 1. ***Continued***

Case No.	Phenotype	Karyotype	Publication
DGAP100	Mental retardation, severe psychomotor delay, mild ventriculomegaly, failure to thrive, no speech, no ambulation, cleft palate, impaired hearing, bilateral optic nerve hypoplasia, severe myopia, hypoglycemia, mild pectus excavatum, gray teeth with caries	46,X,t(X;5)(p11.3;q35.2)dn	
DGAP101	Severe mental retardation, no speech, mild dysmorphism, clinodactyly, mild hirsutism	46,XY,inv(5)(q13q15)dn	
DGAP103	Extreme somatic overgrowth, advanced endochondral bone and dental ages, a cerebellar tumor, multiple lipomas	46,XY,inv(12)(p11.22q14.3)dn	[3]
DGAP104	Congenital hydrocephalus, abnormal corpus callosum, periventricular calcifications, sacral anomaly, hypoplastic kidneys	46,XX,t(1;20)(p31.3;q13.31)dn	[23]
DGAP105	Aortic coarctation; bicuspid aortic valve; bilateral cryptorchidism and primary hypospadias; inguinal hernia; widely spaced nipples; short neck; four hair whorls (three posterior and one anterior); down-slanting palpebral fissures; bilateral epicanthal folds; broad nose; smooth philtrum; thin vermilion border; low-set and posteriorly rotated ears with simplified, thickened helices; mild hypertelorism and strabismus; developmental delay	46,XY,t(1;5)(p35.3;q31.3)dn	
DGAP106	Developmental delay, self-injurious actions and agitation, growth retardation, strabismus, ptosis, normal MRI	46,XX,t(3;5)(q27;q31.1), t(11;13)(p15.3;q14.1)dn	
DGAP107	Visual defects, limb defects, urinary tract abnormalities, learning disabilities, genital anomalies, neurological and behavioral defects	46,XY,t(Y;3)(p11.2;p12.3)dn	[20]
DGAP112	Microcephaly, advanced bone age and secondary craniosynostosis, developmental delay, flat nasal bridge, epicanthal folds, strabismus, short philtrum, thin upper lip, two café-au-lait spots, 2nd toes overlap 3rd toes bilaterally, small labia majora, extra creases on right hand, wide thumbs and halluces	46,XX,t(3;12)(q11.2;q14.1), del(12)(q14q14)dn	
DGAP121	Feeding problems at birth, malformed left ear lobe, epicanthal folds, learning problems, mild hypotonia, mild resolved scoliosis	46,XX,t(5;13)(q15;q32)dn	
DGAP122	Epicanthal folds; hypertelorism; frontal and posterior cowlick; coarse hair; area of alopecia; history of patchy, intermittent hair loss; partially attached pinnae; mild micrognathia; mild pectus excavatum; soft systolic murmur with normal echocardiogram; developmental delay; renal insufficiency caused by grade II-III hydronephrosis	46,XY,t(1;9;5)(1pter→1q32::9p22-24::5q15→5qter;9pter→9p24::9p22→9qter;5pter→5q15::1q32→1qter)dn*	
DGAP123	Autism	46,XX,ins(16;2)(q13;p16.1p16.3) pat.ish ins(16;2)(wcp2+;wcp2+)	[36]
DGAP127	Failure to thrive; feeding problems; growth retardation; unexplained weight loss; brachycephaly; flat mid-face; pointed chin; broad, prominent forehead; deeply set eyes; small mouth; frequent episodes of abdominal pain; some difficulties with reflux; kidney stones; developing contractures and spasticity of the ankles, knees, elbows and shoulders; severe developmental delay; very poor eye contact/interaction; self-stimulating episodes; episodic discomfort and agitation with no apparent cause; seizures; muscle biopsy demonstrated partial complex III deficiency	46,X,t(X;5)(q26.2;q14.3)dn	
DGAP128	Macrocephaly, significant developmental delay, seizures and cerebral atrophy	46,XX,t(1;3)(q32.1;q25.2)dn	[35]
DGAP137	Mild mental retardation, pigment abnormality, VSD, conductive hearing loss, abnormal thyroid function tests, right eye poor visual acuity (small pit in right optic nerve), bilateral optic nerve colobomas, MRI shows 1.5 cm mass behind right globe (no enhancement), bulbous great toes with convex toenails, ligamentous laxity, easy bruising	46,XX,der(6)t(6;13)(q23.3;q22) inv(6)(p21.3q15),der(13)t(6;13)(q23.3;q22)dn	
DGAP139	Developmental delay; hypotonia; dolicocephaly; frontal upsweep; synophrys; long, straight eyelashes; small nares; pronounced philtral creases; small mouth; flat hemangiomas on back of neck; pectus excavatum; joint hyperextensibility; feet have increased secondary creases on both soles; hands have a right Sydney line	46,XY,t(7;13)(p15.3;q14.1)dn	
DGAP151	Cleft lip and palate	46,XX,t(2;8)(q33.1;q24.3)dn	[19]
DGAP157	Global developmental delay, bilateral inguinal hernia, spina bifida occulta, mild dysmorphic features	46,XY,t(3;10)(p12.3-14;q21)dn	

DGAP159	Growth retardation, brachydactyly, bilateral syndactyly of 2nd and 3rd toes, micrognathia, low-set ears, hypertelorism and single palmar crease, developmental delay, no oral language, some autistic and ADD behaviors, abnormal brain CT (5 months of age), moderate to severe bilateral conductive hearing loss, hypo and hypersensitive to different tactile stimulation, trouble focusing eyes on close objects	46,XY,t(8;10)(q13;p14)dn	
DGAP166	Seizure disorder, developmental delay, microcephaly, bilateral epicanthal folds, nose upturned with a thin upper lip and upturned corners of the mouth, very mild micrognathia	46,XX,inv(2)(p22.1q24.3)dn	
DGAP167	Mild developmental delay, vertical talus (rocker-bottom foot deformity), hypotonia	46,XX,inv(18)(q11.2q23)dn	
DGAP169	Failure to thrive, feeding problems, growth retardation, bilateral microtia with profound sensorineural deafness, fused incus and malleus, incus with absent short process, bilateral Mondini malformation, abnormal cochlear turn, malformation of the semicircular canals, micrognathia, anteriorly displaced larynx, small right kidney with renal cortical thinning, borderline wide interpedicular distance of C-spine (18 mm C7, 15–16 mm C5), developmental delay, abnormal hair distribution with high forehead, benign precocious thelarche at 9 months that resolved by 15 months	46,XX,inv(5)(q15q33.2), del(5)(q15)dn	
DGAP173	Mild developmental delay; major depression; generalized anxiety; sleep apnea; self-injurious behaviors; agitation; tantrums; overgrowth; male-pattern hirsutism; amenorrhea; impaired glucose tolerance; hypercholesterolemia and hypertriglyceridemia; elevated testosterone; deep voice; history of one seizure at 2 yr of age; eczema; acanthosis nigricans; moles and skin tags; bilateral epicanthal folds; small nose; complex malocclusion; short, hyperkeratotic palms; 5th finger brachydactyly and clinodactyly; right elbow extension limitation; hypoplastic toenails; short feet	46,XX,t(2;11)(q11.2;p13)dn	
DGAP174	Overgrowth, right-sided hemihypertrophy, small apical VSD and current heart murmur, metopic craniosynostosis and hydrocephalus, Arnold Chiari II malformation, agenesis of the corpus callosum, dysplasia of the left temporal lobe, scoliosis, developmental delay, attention deficit hyperactivity disorder, increased red blood cell size, asthma and seasonal allergies, left inguinal hernia that was surgically repaired, bilateral epicanthal folds, slight occasional esotropia, high-arched palate, left-sided head tilt, unsteady gait and uncoordinated movements with decreased balance	46,XY,t(1;3)(p31.3;q25.1), del(1)(p31.3p32.1)dn	[23]
DGAP190	Developmental delay, infantile spasms	46,XX,t(X;8)(p22;p21)dn	
DGAP200	PDD-NOS, ADHD, conduct disorder with early onset, intermittent explosive disorder, obesity	46,XY,t(1;2)(q31.3;p16.3)dn	[36]

*indicates reported karyotype; FISH analyses suggest additional rearrangements that were not characterized further.

*Correspondence: cmorton@partners.org
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