Supplemental Data

Breakpoint Mapping and Array CGH in Translocations: Comparison of a Phenotypically Normal and an Abnormal Cohort

Julia Baptista, Catherine Mercer, Elena Prigmore, Susan M. Gribble, Nigel P. Carter, Viv Maloney, N. Simon Thomas, Patricia A. Jacobs, and John A. Crolla

Table S1. Phenotypic Details of 18 Clinically Normal Individuals

	Case 1A	Case 1B
Age at Assessment	53 years	35 years
Birth History	No data	Term
Medical History	Intermittent bilateral hip and knee pain from age 6 years Tonsillectomy in childhood Left hip replacement age 52 years Right hip replacement pending Hypercholesterolaemia not requiring treatment Allergic to aspirin Teeth all lost secondary to trauma, e.g. rugby "Soft" secondary dentition?	Perthe's disease, aged 8 years Road traffic accident, aged 14—sustained metatarsal fracture Age 20—onset of back pain and sciatica Femoral osteotomy aged 25 years, leg length discrepancy of 2 cm diagnosed subsequently Cluster of fainting episodes in late 20s, no causative diagnosis made
Developmental History	No history of developmental delay Academic achievement consistent for family	No history of developmental delay Academic achievement consistent for family
Vision and Hearing	No hearing difficulties Myopic since teenage years	No hearing difficulties No visual difficulties
Height	173 cm	162.5 cm
Percentile	25^{th}–50^{th}	25th–50^{th}
Est. Height from Parental Heights	179 cm	169 cm
Occipital-Frontal Circumference	57 cm	56 cm
Percentile	25^{th}–50^{th}	50^{th}–75^{th}
General Examination	Faded strawberry naevus, right thigh	Unremarkable
Neurological Examination	Normal tone, power, and coordination in all four limbs	Normal tone, power and coordination in all four limbs
Dysmorphic Features	None	None
Family History	None	None

	Case 1C	Case 2A
Age at Assessment	62 years	46 years
Birth History	No data	No data
Medical History	No health problems in childhood Cervical polyps Age 38, episode of collapse, secondary to anaemia. No overt loss and no cause found. No further episodes. Arthritis in small joints	No health problems in childhood Episode of epigastric pain aged 45 years, no diagnosis made but splenic cyst found incidentally Prolactinoma diagnosed at 43 years, requiring medical treatment with Cabergoline
Developmental History	No history of developmental delay Academic achievement consistent for family	No history of developmental delay Academic achievement consistent for family
Vision and Hearing	No hearing difficulties Amblyopia and reduced vision in left eye	No hearing difficulties No visual difficulties. Visual fields normal.
Height	160 cm	160 cm
Percentile	25^{th}–50^{th}	25^{th}–50^{th}
Est. Height from Parental Heights	163 cm	No data

Occipital-Frontal Circumference	57 cm	55 cm
Percentile	75^{th}–91^{st}	25^{th}–50^{th}
General Examination	Unremarkable	Unremarkable
Neurological examination	Normal tone, power and coordination in all four limbs	Normal tone, power and coordination in all four limbs
Dysmorphic Features	None	None
Family History	None	None

	Case 2B	**Case 2C**
Age at Assessment	35 years	40 years
Birth History	Term	No data
Medical History	Nasal polyps removed aged 12 years Unilateral oophrectomy due to ovarian cyst, aged 21 years Recurrent knee dislocations, arthroscopy aged 22 years. ×1 miscarriage, ×1 TOP for multiple fetal anomalies, further ×6 miscarriages	Tonsillectomy aged 6 years No other significant history in childhood Single episode of chest pain during coitus. No cause found; diagnosed as musculo-skeletal
Developmental History	No history of developmental delay Developed speech aged 3 years Academic achievement consistent for family	No history of developmental delay Slow learning to read, educated in special needs class until age 10 years Progress then rapid, completed university education
Vision and Hearing	No hearing difficulties Borderline hypermetropia	No hearing difficulties No visual difficulties
Height	150 cm	170 cm
Percentile	0.4^{th}–2^{nd}	9^{th}–25^{th}
Est. Height from Parental Heights	158 cm	167.5 cm
Occipital-Frontal Circumference	51.5 cm	57 cm
Percentile	$< 0.4^{th}$	25^{th}–50^{th}
General Examination	Dental crowding, gum hypertrophy and gum inflammation	Single large naevus on right thigh No other skin markings
Neurological examination	Mild intention tremor in both hands Mildly impaired balance on heel-to-toe walking	Normal tone, power and coordination in all four limbs
Dysmorphic Features	High arched palate	None
Family History	Daughter with same translocation, developmentally normal Single palmar crease	None

	Case 2D	**Case 2E**
Age at Assessment	55 years	29 years
Birth History	No data	No data
Medical History	Recurrent tonsillitis Hepatitis A aged 25 years Cervical intraepithelia neoplasia, treated by colposcopy and ablation ×3 miscarriages Pneumonia aged 44 years	Molar pregnancy aged 23 years, presented at 6 weeks gestation. ×3 miscarriages
Developmental History	No history of developmental delay Slow learning to read, possibly dyslexic Eventual academic achievement consistent for family	No history of developmental delay Walked at less than 1 year
Vision and Hearing	No hearing difficulties Myopic	No hearing difficulties No visual difficulties
Height	162.5 cm	155 cm
Percentile	25^{th}–50^{th}	9^{th}
Est. Height from Parental Heights	165 cm	159 cm

Occipital-Frontal Circumference	56 cm	55 cm
Percentile	50^{th}–75^{th}	25^{th}–50^{th}
General Examination	Two small naevi on left upper arm	Unremarkable
Neurological examination	Normal tone, power and coordination in all four limbs	Normal tone, power and coordination in all four limbs
Dysmorphic Features	None	Small ears, right ear overfolded
Family History	Daughter with same balanced translocation has normal phenotype	Overfolded ear found in two other family members

	Case 2F	**Case 2G**
Age at Assessment	59 years	58 years
Birth History	Term	Term
Medical History	Limited past medical history No hospital admissions other than to treat numerous fractures, all due to horse riding accidents. Single episode of "blackout" aged 21 years. Depression aged 58 years. Partner ×11 miscarriages	Ear polyps diagnosed aged 5 years.. Road-traffic accident aged 19, causing multiple fractures. Low sperm count; conceived after 10 years. Deep vein thrombosis, secondary to trauma aged 40 years.
Developmental History	No data Unable to read, poor writing skills, but academic achievement consistent for family	No history of developmental delay Mild dyslexia
Vision and Hearing	No hearing difficulties Hypermetropia, recent onset. Previously no difficulties	Mild hearing impairment secondary to loud noise exposure No visual difficulties
Height	162.5 cm	188 cm
Percentile	0.4^{th}–2^{nd}	91^{st}–98^{th}
Est. Height from Parental Heights	154 cm	No data
Occipital-Frontal Circumference	55 cm	59 cm
Percentile	9^{th}	75^{th}–91^{st}
General Examination	Unremarkable	Unremarkable
Neurological Examination	Normal tone, power and coordination in all four limbs	Normal tone, power and coordination in all four limbs
Dysmorphic Features	None	No dysmorphic facial features Feet—low arches
Family History	None	Son with same translocation, with developmental delay and epilepsy

	Case 2H	**Case 3A**
Age at Assessment	33 years	41 years
Birth History	Born at 39/40 weeks, 3.5 kg	No data
Medical History	No childhood medical problems Left knee arthroscopy and trimming of cartilage. Right knee recurrent bursitis	Asthma diagnosed aged 5 years. Mild asthma only as an adult Childhood eczema ×7 first trimester miscarriages, originally thought due to cervical incompetence Breast Cancer, aged 37 years Metastatic disease diagnosed aged 40 years
Developmental History	No history of developmental delay Academic achievement consistent for family	No history of developmental delay Academic achievement consistent with family
Vision and Hearing	No hearing difficulties Right amblyopia, under treated, subsequent poor vision in right eye	No hearing difficulties No visual difficulties
Height	185 cm	152.5 cm
Percentile	75^{th}–91^{st}	2^{nd}–9^{th}

Est. Height from Parental Heights	174 cm	168 cm
Occipital-Frontal Circumference	60.5 cm	54 cm
Percentile	91^{st}–98^{th}	9^{th}–25^{th}
General Examination	Raised body mass index	Cachetic
Neurological Examination	Normal tone, power and coordination in all four limbs	Bradykinesia, consistent with terminal disease No focal neurological signs
Dysmorphic Features	High arched palate No dysmorphic facial features Feet—low arches	None
Family History	Two sons with Beckwith-Wiedemann syndrome, due to duplication of 11p	None

	Case 3B	**Case 3C**
Age at assessment	35 years	36 years
Birth History	Term	No data
Medical History	No medical problems as a child Breast reduction surgery, requiring repeat procedures ×2 miscarriages	Recurrent chest infections as a child Mild asthma Heart murmur diagnosed in childhood; resolved spontaneously ×3 miscarriages
Developmental History	No history of developmental delay	No history of developmental delay Speech and language mildly delayed Academic achievement consistent for family
Vision and Hearing	No hearing difficulties No visual difficulties	No hearing difficulties No visual difficulties
Height	162.5 cm	162.5 cm
Percentile	25^{th}–50^{th}	25^{th}–50^{th}
Est. Height from Parental Heights	No data	164 cm
Occipital-Frontal Circumference	56 cm	57 cm
Percentile	50th–75th	75^{th}–91^{st}
General Examination	Single café au lait patch on left aspect of neck	Area of freckling on right forearm, 2 cm × 3 cm
Neurological Examination	High arched palate Normal tone, power and coordination in all four limbs	Normal tone, power and coordination in all four limbs
Dysmorphic Features	None	None
Family History	Son with unbalanced translocation, multiple medical problems and global delay	Daughter with developmental delay and epilepsy, normal karyotype, diagnosis: cerebral palsy

	Case 3D	**Case 3E**
Age at Assessment	38 years	48 years
Birth History	Term	No data
Medical History	Right orchidopexy aged 9 years Renal stone aged 11 years, no underlying cause found Ruptured left side prepatellar bursa aged 35 years.	Rheumatic fever aged 4 years Tonsillectomy aged 5 years Borderline hypertension
Developmental History	No history of developmental delay Academic achievement consistent for family	Walked aged 1 year Talked “early” Academic achievement consistent with family
Vision and Hearing	No hearing difficulties Right strabismus in infancy, corrected at 2 years	No hearing difficulties No visual difficulties
Height	178 cm	162.5 cm
Percentile	50^{th}–75^{th}	25^{th}–50^{th}
Est. Height from Parental Heights	176.5 cm	168 cm
Occipital-Frontal Circumference	58 cm	57 cm

Percentile	50^{th}–75^{th}	75^{th}–91^{st}
General Examination	Unremarkable	Unremarkable
Neurological Examination	Normal tone and power Mild dysdiadodyskinesis; lower limb coordination normal	Normal tone, power and coordination in all four limbs
Dysmorphic Features	Ears—prominent crus	Feet—large for height, flat arches
Family History	None	Daughter with unbalanced product of patient's translocation. Mild Asperger syndrome, memory/concentration difficulties

	Case 3F	**Case 3G**
Age at Assessment	57 years	41 years
Birth History	No data	No data
Medical History	No medical problems during childhood ×1 miscarriage Umbilical hernia, repaired aged 55 years	Tonsillectomy aged 13 years 34 years: severe road traffic accident, involving fractured femur and wrist. Residual leg length discrepancy Blackout during admission. Embolic episode? Following dental work, has been commented that teeth are very dense ×4 miscarriages Breast cyst
Developmental History	No history of developmental delay Academic achievement consistent for family	No history of developmental delay Academic achievement consistent for family
Vision and Hearing	Mild hearing loss, age related Myopia, prescription –5, required glasses since 8 years	No hearing difficulties No visual difficulties
Height	157.5 cm	160 cm
Percentile	9^{th}–25^{th}	25^{th}–50^{th}
Est. Height from Parental Heights	160 cm	161.5 cm
Occipital-Frontal Circumference	56.5 cm	55.2 cm
Percentile	75^{th}–91^{st}	25^{th}–50^{th}
General Examination	Unremarkable	Small strawberry naevus on breast
Neurological Examination	Normal tone, power and coordination in all four limbs	Normal tone, power and coordination in all four limbs
Dysmorphic Features	None	Ears—large upper part of pinnae, small ear lobes, normally rotated Feet—flat arches
Family History	None	Son has normal phenotype and same balanced translocation

Table S2. Phenotypic Details of the 14 Clinically Affected Patients

	Case 16	**Case 20**
Age at Assessment	34 years	32 years
Birth History	Term. Born in good condition	38/40. Birth weight 2.18 kg. 2^{nd} percentile
Medical History	Absence seizures from 4 years. Age 6 years grand mal seizures, diagnosed as temporal lobe epilepsy Psychiatric history, beginning at 11 years. Obsessional behaviour and anxiety requiring prolonged psychiatric admission Obesity, treated with gastric bypass	Quiet and hypotonic from birth Ventilated for 1/7, required nasogastric feeds for 4/7. Remained hypotonic until ambulant. Menorrhagia. Migraines throughout teenage years
Developmental History	Sat at 6 months, walked at 18 months. Speech development normal. No concerns until age 11 years. Adult IQ 74	Sat at 6 months, walked at 14 months. Speech development slow, main difficulty with understanding. Attended mainstream school with support
Vision and Hearing	Mild hearing impairment. No visual difficulties	No hearing difficulties. Amblyopia

Height	162.5 cm	157.5 cm
Percentile	25^{th}–50^{th}	9^{th}–25^{th}
Est. Height from Parental Heights	158 cm	158 cm
Occipital-Frontal Circumference	58 cm	55 cm
Percentile	98^{th}	25^{th}–50^{th}
General Examination	Flat affect with monotonal voice, high body mass index	Slow, slightly slurred speech. Quiet, shy
Neurological Examination	Bradykinesia. Normal tone and power in all four limbs Poor coordination and heel-to-toe walk. Normal eye movements.	Normal tone and power in all four limbs. Poor coordination. Unable to heel-to-toe walk. Poor upper limb coordination.
Dysmorphic Features	Downslanting palpebral fissures	Coarse facial features. Dark, curly hair. Single palmar crease on left hand
Family History	Brother with temporal lobe epilepsy and normal karyotype	Sister with anorexia nervosa

	Case 43	**Case 45**
Age at Assessment	3 months	32 years
Birth History	No data	No data
Medical History	Isolated truncus arteriosus	No medical problems as a child. Menarche aged 12 years. Menses initially erratic and then regular. Amenorrhoea from age 31 years. No hot flushes or other menopausal symptoms. Gonadotrophin levels high consistent with premature menopause
Developmental History	Normal development	No delay known in early milestones. Attended mainstream school
Vision and Hearing	No data	No data
Height	No data	157 cm
Percentile	No data	9^{th}–25^{th}
Est. Height from Parental Heights	No data	No data
Occipital-Frontal Circumference	No data	No data
Percentile	No data	No data
General Examination	No data	Unremarkable
Neurological Examination	No data	Neurology grossly intact
Dysmorphic Features	None	None
Family History	No data	No data

	Case 48	**Case 49**
Age at Assessment	15 years	21 years
Birth History	No data	No data
Medical History	No data	Isolated cleft palate repaired at 6 months. Two operations for scoliosis
Developmental History	Severe developmental delay	No speech, but good communication with British sign language
Vision and Hearing	No data	No data No data

Height	No data	No data
Percentile	No data	No data
Est. Height from Parental Heights	No data	No data
Occipital-Frontal Circumference	No data	No data
Percentile	No data	No data
General Examination	No data	Very sociable but with significant behavioural abnormalities
Neurological Examination	No data	No data
Dysmorphic Features	Dysmorphic	Long thin fingers with hypoplastic distal phalanges. Long face with a pointed chin and a high anterior hairline with a cowlick. Coarse features, including a long nose.
Family History	No data	No data

	Case 50	**Case 51**
Age at Assessment	30 years	6 years
Birth History	No data	Term. 2.86 kg Feeding difficulties, wouldn't breastfeed
Medical History	Menarche aged 14 years. Menses at one year intervals. Aged 19 years took combined oral contraceptive pill for 9 years; on discontinuation, no resumption of menstruation, associated with high gonadotrophins	Autistic Spectrum Disorder and Attention Deficit Hyperactivity Disorder aged 6 years Sleeps poorly, requiring melatonin Shows mannerisms and repetitive behaviours, self injurious behaviour No seizures
Developmental History	No delay known in early milestones	Rolled at 1 year, sat at approximately 1 year, walked at 19 months No language until 4 years Severe language delay Attends special needs school
Vision and Hearing	No hearing difficulties No visual difficulties	No hearing difficulties No visual difficulties
Height	166 cm	131.6 cm
Percentile	50^{th}–75^{th}	75^{th}–91^{st}
Est. Height from Parental Heights	159 cm	Tall for family
Occipital-Frontal Circumference	56 cm	50.5 cm
Centile	50^{th}–75^{th}	2^{nd}
General Examination	Slight build, long fingers	Tall stature 1 café au lait patch on left back
Neurological Examination	Neurology grossly normal	Neurology grossly normal
Dysmorphic Features	No dysmorphic features	Large, upslanting eyes Well defined philtrum Mild fifth finger clinodactyly
Family History	Mother had very irregular menses, menarche at 15 years, menopause at 50 years, normal karyotype	Brother with 47XYY in 90% of cells and marker chromosome 22

	Case 52	**Case 53**
Age at Assessment	13 years	7 years
Birth History	No data	41/40. 3.83 kg. Born in good condition
Medical History	Respiratory problems and vocal cord palsy, which required tracheostomy. Scoliosis (60%) required surgery. Learning difficulties	Failed to fix with eyes at 3/12 8/12 hypotonia identified. Episode of loss of use of left upper and lower limbs, aged 3 years. CT scan normal, recovery made but slight weakness

	Normal growth rate. Short stature.	remained. Seizure at 8/12 Febrile fit? Initial MRI brain—delayed myelination. Repeat scan aged 6 years, complete myelination
Developmental History	Delayed speech	Smiled at 6 weeks, rolled at 6 months, sat at 18 months. Able to walk with support. No speech, understands limited number of words
Vision and Hearing	No data	Hearing "excellent" Hyperacusis? Left strabismus
Height	No data	No data
Percentile	No data	No data
Est. Height from Parental Heights	No data	No data
Occipital-Frontal Circumference	No data	52 cm
Percentile	2^{nd}	25^{th}
General Examination	Asymmetry more marked in the feet	Left strabismus, quiet
Neurological Examination	No data	Hypotonic, spontaneous movements of all four limbs. Deep tendon reflexes present, plantars downgoing
Dysmorphic Features	Slight facial dysmorphism	No dysmorphic facial features Slender fingers
Family History	No data	No data

	Case 54	**Case 55**
Age at Assessment	11years	42 years
Birth History	Term. 3.8 kg. Born in good condition	No data
Medical History	Coarctation of aorta diagnosed at 8 weeks. Associated with cardiomegaly, pneumothorax, and pneumonia ×2 further pneumothoraces in 1st year. Repeated chest infections. Undescended testes, orchidopexy at 3 years. Behavioural difficulties, on autistic spectrum	No childhood medical problems Allergies, including hayfever Gastro-oesophageal reflux, diet controlled Gum disease and poor dentition Low sperm count Partner had "multiple" miscarriages
Developmental History	Obsessional tendencies but very sociable. Hypercholesterolaemia, diet controlled. Smiled at 10 weeks, sat at 9 months, walked at 17 months. Speech delay. Attends mainstream school, mild learning difficulties	No known delay in early milestones Attended mainstream school Academic achievement consistent with family
Vision and Hearing	No hearing difficulties Left strabismus, corrected at 5 years	No hearing difficulties No visual difficulties
Height	No data	175 cm
Percentile	No data	25^{th}–50^{th}
Est. Height from Parental Heights	Tall for family	176 cm
Occipital-Frontal Circumference	57 cm	60.5 cm
Percentile	98^{th}–99.6^{th}	91^{st}–98^{th} (Brother also has a large head)
General Examination	Tall stature Left strabismus	Obese
Neurological Examination	Normal tone and power in all four limbs Mild ataxia demonstrated by reduced ability to heel-to-toe walk	Normal, tone power and coordination in all four limbs. High palate
Dysmorphic Features	Ears—large ear lobes, flat pinnae Hands—brachydactyly, mild joint hypermobility Feet—wide with short toes, low arches Pectus excavatum	Hyperteloric Long, slim fingers

Family History	Father has hypercholesterolaemia	No data

	Case 56	**Case 57**
Age at Assessment	39 years	43 years
Birth History	No data	41/40. Birth weight 2.55 kg. 2nd percentile. Quiet on delivery, required nasogastric feeding for 3 weeks
Medical History	No childhood medical problems Infertility, with very low spem count, mainly immotile	Epilepsy diagnosed at 2 years, grand mal seizures. Had a seizure every few months during childhood. No episodes of status epilepticus. Hypertension, diagnosed at 40 years. Hypercholesterolaemia. Joint laxity, knee dislocation requiring surgery
Developmental History	No known delay in early milestones Academic achievement consistent with family Difficulties in visuo-spatial assessment; for example, unable to drive	Sat at around 8 months, walked at 16 months. Speech development normal. Attended mainstream school. Paid unskilled employment, lives independently. Academic achievement less than unaffected family members
Vision and Hearing	No hearing difficulties No visual difficulties	No hearing difficulties. No visual difficulties
Height	180 cm	160 cm
Percentile	50th—75th	0.4th–2nd
Est. Height from Parental Heights	178 cm	171.5 cm
Occipital-Frontal Circumference	57.5 cm	No data
Percentile	50th—75th	No data
General Examination		Speech slow but not dysarthric. Normal palate Dental crowding
Neurological Examination	Normal tone, power and co-ordination in all four limbs	Normal tone and power in all four limbs. Poor co-ordination of upper and lower limbs. Unsteady gait, unable to heel-toe walk. Normal eye movements
Dysmorphic Features	No dysmorphic facial features. Feet- very flat arches. Pectus carinatum	No dysmorphic facial features
Family History	No data	Brother with normal karyotype and intrauterine growth retardation, stellate cataract strabismus, short stature, learning difficulties and depression. No facial dysmorphic features

Table S3. Genes Mapped to the Deleted Regions in Cases 20, 57, 52, and 53

Case	Chromosome	Gene Symbol	Gene Name
20	5q12.1	*KIF2A*	kinesin heavy chain member 2A
20	5q12.1	*DIMT1L*	DIM1 dimethyladenosine transferase 1-like (S. cerevisiae)
20	5q12.1	*IPO11*	importin 11
20	5q12.1	*ISCA1L*	iron-sulfur cluster assembly 1 homolog (S. cerevisiae)-like
20	5q12.1	*HTR1A*	5-hydroxytryptamine (serotonin) receptor 1A
20	5q12.1	*RNF180*	RING finger protein 180
52	4q13.3	SLC4A4	solute carrier family 4, sodium bicarbonate cotransporter, member 4
52	4q13.3	*GC*	group-specific component (vitamin D binding protein)
52	4q13.3	*NPFFR2*	neuropeptide FF receptor 2
52	4q13.3	*ADAMTS3*	metallopeptidase with thrombospondin type 1 motif, 3
52	4q13.3	*COX18*	cytochrome c oxidase assembly homolog (S. cerevisiae)
52	4q13.3	*ANKRD17*	ankyrin repeat domain 17
53	4q13.3	*PF4V*	platelet factor 4 variant 1
53	4q13.3	*CXCL1*	chemokine (C-X-C motif) ligand 1 (melanoma growth stimulating activity, alpha)
53	4q13.3	*PF4*	platelet factor 4 (chemokine (C-X-C motif) ligand 4)
53	4q13.3	*PPBP*	pro-platelet basic protein (chemokine (C-X-C motif) ligand 7)
53	4q13.3	*CXCL5*	chemokine (C-X-C motif) ligand 5
53	4q13.3	*CXCL3*	chemokine (C-X-C motif) ligand 3
53	4q13.3	*PPBPL2*	pro-platelet basic protein-like 2
53	4q13.3	*CXCL2*	chemokine (C-X-C motif) ligand 2
53	4q13.3	*MTHFD2L*	methylenetetrahydrofolate dehydrogenase (NADP+ dependent) 2-like
53	4q13.3	*EPGN*	epithelial mitogen homolog (mouse)
53	4q13.3	*EREG*	epiregulin
53	4q13.3	*AREG*	amphiregulin (schwannoma-derived growth factor)
53	4q13.3	*BTC*	betacellulin
53	4q13.3	*PARM1*	Protein PARM-1 precursor.
53	4q13.3	*RCHY1*	ring finger and CHY zinc finger domain containing 1
53	4q13.3	*THAP6*	THAP domain containing 6
53	4q13.3	*CDKL2*	cyclin-dependent kinase-like 2 (CDC2-related kinase)
53	4q13.3	*G3BP2*	GTPase activating protein (SH3 domain) binding protein 2
53	4q13.3	*USO1*	USO1 homolog, vesicle docking protein (yeast)
53	4q13.3	*PPEF2*	protein phosphatase, EF-hand calcium binding domain 2
53	4q13.3	*ASAHL*	N-acylsphingosine amidohydrolase (acid ceramidase)-like
57	4q32.1	*MAP9*	microtubule-associated protein 9
57	4q32.1	*GUCY1A3*	guanylate cyclase 1, soluble, alpha 3
57	4q32.1	*GUCY1B3*	guanylate cyclase 1, soluble, beta 3
57	4q32.1	*ACCN5*	amiloride-sensitive cation channel 5, intestinal
57	4q32.1	*TDO2*	tryptophan 2,3-dioxygenase
57	4q32.1	*CTSO*	cathepsin O
57	4q32.1	*PDGFC*	platelet derived growth factor C
57	4q32.1	*GLRB*	glycine receptor, beta
57	4q32.1	*GRIA2*	glutamate receptor, ionotropic, AMPA 2