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Supplemental Data

WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome

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Supplemental note: Case Reports

***WNT5A* CASE SUMMARIES**

BAB9138

BAB9138 is a seven year old boy born to parents with possible consanguinity. He has short stature (-4.25 SD), telecanthus, hypertelorism, frontal bossing, prominent eyes, anteverted nares, wide and depressed nasal bridge, midface hypoplasia, smooth philtrum, wide mouth, bilobed tongue, gingival hyperplasia, microretrognatia, and low-set ears. He has significant mesomelic limb shortening, and fingers and toes are very broad and short and nails are dysplastic. He had prior surgery to remove Y-shaped duplication of bilateral thumbs and great toes. He has a buried penis, cryptorchidism and sacral dimple. Radiographs revealed hemivertebrae (T6, T7, T13). Early motor milestones were delayed but present cognitive development is normal.

BAB9131

Subject BAB9131 is a 17 year old female. She was born at term with average birth parameters after an uncomplicated pregnancy to unrelated parents. She presented in infancy with slow linear growth marked by acromesomelia, hypertelorism and a prominent forehead. A clinical diagnosis of Robinow syndrome was made. By 18 months of age, height/length was 71.5 cm (-2.89 SD), weight was 9.15 kg (-1.97 SD), and high hyperopia was detected. She had normal motor, language and social development, delayed primary and secondary tooth eruption and dental crowding. By 8-3/4 years of age, height was 117 cm (-2.49 SD), weight was 25.8 kg (33%) and radiographs of the hands showed bifid tufts of the distal thumb. Research sequencing of *ROR2* was unrevealing though Robinow syndrome remained her clinical diagnosis. At 10 years of age she underwent bilateral distal tibial hemiepiphysiodesis and osteotomies for bilateral valgus ankle deformity and pain. This procedure improved her ankle pain, but she continued to have mild, diffuse pain of her hips, knees, elbows and shoulders, managed successfully with stretching and exercise. Menarche occurred at 12 years of age and was irregular until treated with OCPs. Laboratory evaluation to evaluate her irregular menses and hypertrichosis were normal. Multiple teeth were extracted and orthodontia implemented to align her teeth. In her mid teens, she began treatment for major depressive disorder. At her last encounter at 17 years of age, she was healthy and physically active without major medical concerns. Height was 144.5 cm (-2.85 SD) and weight was 48.5 kg (18%).

XpTER DELETION AND 6qTER DUPLICATION CASE SUMMARY

BAB8836

BAB8836 is a male and the third child of healthy Turkish parents who have consanguineous marriage. His brother and sister are healthy and there is no significant family history. He was born at term by vaginal delivery and was monitored for one week in neonatal period to reduce the levels of newborn bilirubin. At birth dysmorphic features were noted with short stature, large cranium, short neck, blue sclera, hypoplastic lower eyelids, flat nasal bridge, narrow rib cage, pectus carinatum, short arms and legs, and anal stenosis. After his second month, he developed xerosis. Milestones included unsupported sitting at 12-13 months, walking at 2 years old, and one word speaking at about 3-4 years; IQ measured at 8 years of age was 74. Echocardiography was normal and cranial MRI and abdominal ultrasound were also normal. Hemivertebra of T12 was noted radiographically and associated with kyphosis. He has post-natal short stature but weight is normal. Genetic testing revealed NM_0045603 p.R244W (c.730 C>T) (Heterozygote) alteration in the *ROR2* gene. Chromosome analysis was normal. Two large CNVs were observed in exome analysis by XHMM and HMZDelFinder and confirmed by aCGH: A terminal deletion on short arm of chromosome Xpter with approximate breakpoints (hg19) mapping in between chrX: 8,199,541 (min) chrX: 8,321,827 (max) and 6qter with approximate breakpoints mapping in between chr6:157,870,814 (min) and chr6:157,461,178 (max).

SH3PXD2B + *INPPL1* CASE SUMMARY

BAB8759

BAB8759 was a female born to consanguineous parents refered to genetics clinic at day of life 12 because of her facial dysmorphic features including prominent forehead, flat occiput, micrognathia, prominent eyes, hypertelorism, downslanting palpebral fissures, flat nasal bridge, nuchal edema and multiple congenital malformations, including pes equinovarus and atrioventricular septal defect. Chromosome analysis was normal. Her phenotypic features were said to be consistent with Robinow syndrome. This subject is deceased and had a history of recurrent respiratory infections. No radiologic imaging is available.

FZD2 CASE SUMMARIES

5449

Subject 5449 is a 10 year-old boy, second child of unrelated parents of Kosovoan origin. There is no family history of similar clinical findings. He was born at term after an uneventful pregnancy, and had normal birth parameters. He had several congenital anomalies that required surgical intervention, including imperforate anus, bilateral lacrimal duct agenesis, and thyroglossal duct cyst.

He also was noted to have bilateral chorioretinal colobomas, without associated visual impairment. Motor and cognitive development and growth have been normal.

His facial characteristics include a broad forehead, prominent eyes, hypertelorism with wide nasal bridge, hypoplasia of the nasal tip (almost bifid with midline telangiectasia) with anteverted nares, midface hypoplasia, posteriorly rotated and low set ears, and micrognathia. These facial features were considered compatible with Robinow syndrome. Anthropomorphic measurements revealed normal height, weight, and OFC, with also normal growth. Height was 137 cm (45th centile) and OFC 52.5 cm (-0.5 SD) at 10 years-old. Skeletal anomalies include mild mesomelia, bilateral short fibulas and ulna (with ulnar deviated hands and reduced wrist mobility), lower limb asymmetry, and proximally implanted thumbs. He has chronic ankle and wrist pain as a consequence of these ulnar and fibular anomalies. Dental anomalies are mild, consisting of teeth crowding.

Recurrent isolated headaches in the right temporal region prompted a brain CT-scan, which showed a type 1 Chiari malformation.

Chromosome analysis and array-CGH (180k Agilent) were normal. Whole exome sequencing identified a variant of unknown significance in *FZD2*, which matched with other subjects from the present study on GeneMatcher.

BAB8596

BAB8596 is a 6-year-old male born at full term via spontaneous vaginal delivery complicated by placental abruption requiring resuscitation for 13 minutes. His birth weight was 6lbs, 11oz (20^{th} centile) with a birth length of 18.25" (9^{th} centile) and a head circumference of 14" (45^{th} centile). He has had normal developmental milestones. He was referred to genetics for evaluation of skeletal dysplasia and had sequencing of *FGFR3* and *NPR2*. At 18 months of age, bone age was 33 months. Chromosomal microarray revealed a maternally-inherited 1q42.2 duplication that includes two genes that have not been associated with a clinical phenotype. At age 3yr 7mo, head circumference was 51.2 cm (78^{th} centile), height was 85.5 cm (-4 SD). Facial

characteristics include broad forehead, frontal bossing, depressed nasal bridge, anteverted nares, and midface hypoplasia. Brachydactyly and acromesomelic shortening of all extremities are present with a hand length of 9.7cm (<3rd centile) and middle finger length 3.9cm (<3rd centile).

BAB8594-affected mother of BAB8596

BAB8594 is the mother of BAB8596. She is 30-years-old from European origin. Her height is 4'11'' with a family history of short stature. There is no evidence of consanguinity. She has strabismus of her left eye. She has facial characteristics of Robinow syndrome and retrognathia. Hand length is 15.5 cm left and 15.7 cm right (<3rd centile).

BAB8705

BAB8705 is a 5-year-old female who was born at term with a birth weight of 3380 grams (39th centile) and height of 44 cm (1st centile). During the 6th month of pregnancy, ultrasound demonstrated short limbs. Prenatal chromosome and *FGFR3* mutation analyses performed on percutaneous umbilical cord blood sampling were normal. A clinical diagnosis of Robinow syndrome was rendered during the newborn period, based upon facial and limb characteristics. At 5 years of age, height is 94.5 cm (-2.9 SD), OFC is 48.5 cm (3-10th centile), she has facial characteristics of Robinow syndrome, 'V' shaped uvula, gingival hyperplasia, brachydactyly, mesomelia of upper limbs, genital hypoplasia and anteriorly-placed anus.

BAB7987

Subject BAB 7987 is a 15 year old female and was first evaluated at age three. Height is 4´10" (-2.25 SD); she has frontal bossing, midface hypoplasia, facial nevi, prominent eyes, upslanting palpebral fissures, long eyelashes, hypertelorism, short nose with anteverted nares, triangular mouth, accentuated cupids bow, micrognathia and multiple dental abnormalities. She has mesomelic limb shortening, with Madelung deformity, brachydactyly, clinodactyly and broad great toes. She has hypoplastic genitalia and recurrent urinary tract infections.

BAB7988-affected mother of BAB7987

Subject BAB 7988 is a 47 year old woman and is the mother of BAB7987. Her father is reported to be similarly affected, although he was unavailable for study. Her height is 5' (-1.7 SD); she has frontal bossing, midface hypoplasia, facial nevi, prominent eyes, upslanting palpebral fissures, strabismus, long eyelashes, hypertelorism, short nose with anteverted nares, triangular mouth, thin upper lip, cleft soft palate, bifid tongue, and dental abnormalities including supernumerary teeth. She has pectus excavatum, mesomelic limb shortening, with Madelung deformity, brachydactyly, short metacarpals, clinodactyly, broad great toes, and nail dysplasia. Her genitalia are hypoplastic and she has a small bladder.

BAB9254 & BAB9255

These are two siblings of BAB8596 who are 4 and 8 years of age. They have short stature, but are well and do not have facial characteristics of Robinow syndrome (all examined by author V.R.S.).

***NXN* CASE SUMMARIES**

BAB8841

BAB8841 is a 5-year-old female. Prenatal course was normal. Parents are first cousins of Turkish ancestry. At birth an omphalocele was present and was repaired in the first week of life. She has subsequently had two ventral hernias which were also repaired. Other medical concerns include bicuspid aortic valve with dilation of the ascending aorta (max Z-score of 2.0 at 5 months of life), aberrant right coronary artery, growth hormone deficiency (which was documented with a stimulation test and peak hormone level of 6.6 at 60 minutes), recurrent urinary tract infections and possible left kidney duplication, frequent ear infections, mild conductive hearing loss treated with two sets of ear tubes.

Growth parameters were normal at birth, but she developed short stature with relative macrocephaly; at 5 years of age, height was 2%, weight 9% and head circumference 100%. Craniofacial characteristics include a high and broad forehead, hypertrichosis with low anterior hairline, long eyelashes, hypertelorism, broad nasal bridge with anteverted nares, narrow posterior pharynx with absent uvula, gingival hyperplasia, midline groove and anterior notch in the tongue, notch in the upper lip in the midline, low set posteriorly rotated ears with cryptotia of the anterior helical rim. She has

brachydactyly and mesomelic shortening, fifth finger brachydactyly and clinodactyly and broad thumbs and great toes.

Development is delayed. She sat unassisted at 1 year of age, crawled at 18 months, pulled to stand at 30 months and walked at 36 months of age. She finger fed herself at 3 years and could copy a line and circle at 4 years. First word was at 1 year of age. She began to put words together into phrases at 5 years of age. She follows commands and communicates with some words as well as gestures, pointing. She has difficulty with pronunciation.

Chromosome analysis and chromosome microarray in 2011 were normal, and clinical sequencing and deletion/duplication analysis for *WNT5A*, *DVL1*, and *ROR2* were normal.

BAB9844

BAB9844 is a 3 year old girl who was born by caesarean section at 39 weeks gestation for breech presentation. She grew initially around the 25th centile for weight and height, and the 75th for head circumference. There was no concern about her general growth or development. She had an open fontanelle with a prominent metopic region, proptosis, a flat nasal bridge and flat midface with a tented upper lip and hypertrophic gums. She had a small jaw and no cleft. Although her overall length was in the normal range, she had the appearance of short limbs and she had an unusual pattern of hand and foot development with brachydactyly and incurving of her fingers towards the middle finger. There was a wide sandal gap and the same clinodactyly pattern in her feet. She had a nevus sebaceous on her scalp and no other malformation. She had a normal chromosome microarray analysis and Stickler syndrome was excluded clinically and following a panel of gene linked to the diagnosis. *ROR2* sequencing was normal.

BAB9847

BAB9847 is a 6 month old sibling of BAB9844 who was born at 37 weeks gestation by emergency caesarean section. A unilateral cleft lip and palate was detected antenatally, and she was thought to have the same facial profile as her sister. She has underdevelopment of her ear helices bilaterally. She is doing well. These siblings are otherwise remarkably alike in terms of their facial features and limb patterning and therefore exome sequencing was requested looking for a likely new recessive condition.

RAC3 CASE SUMMARY

BAB8740

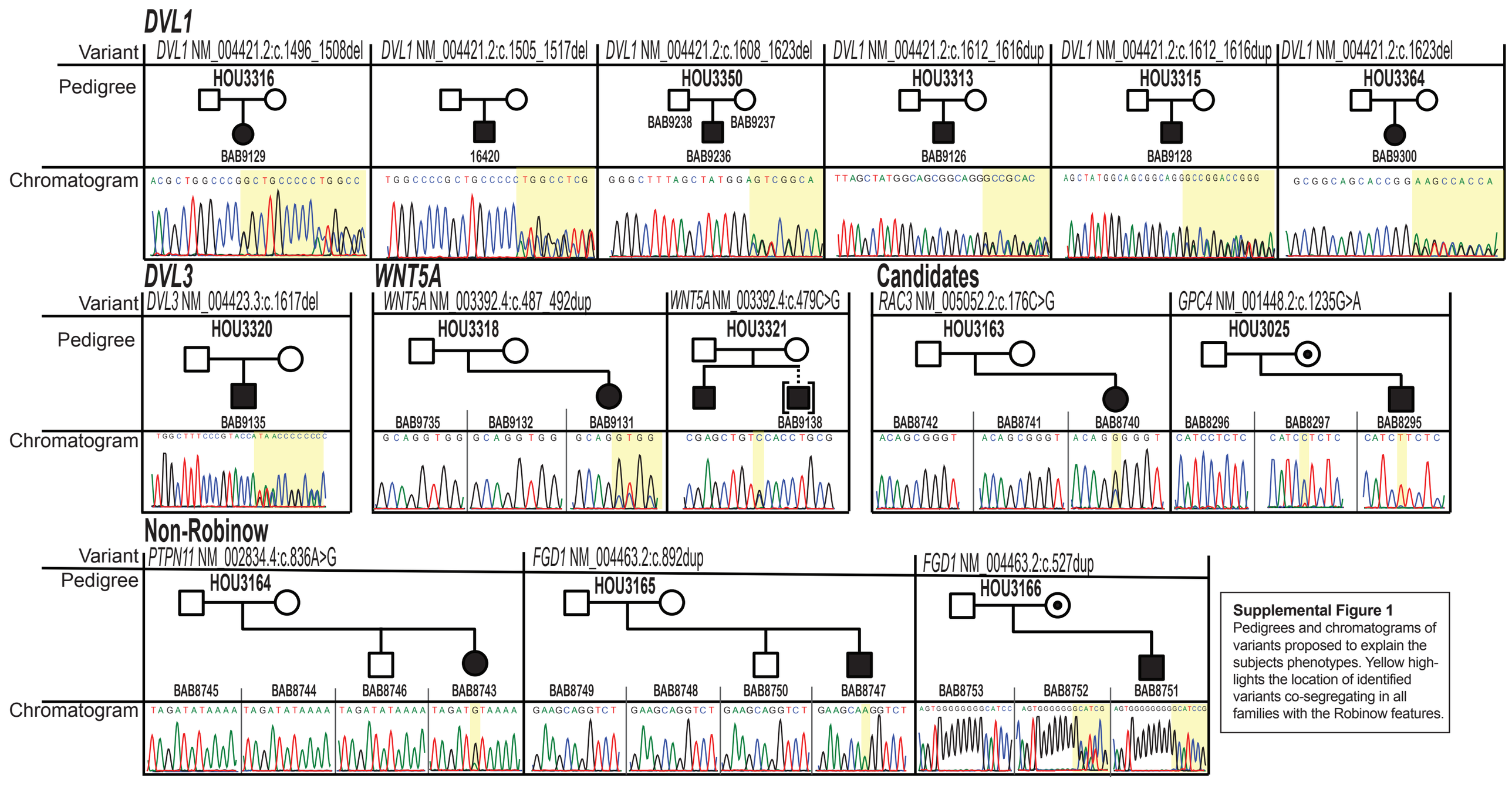
BAB8740 is a 4-year-old female. She is the firstborn of healthy non-consanguineous parents delivered via cesarean section at 38 1/7 weeks gestation after an uneventful pregnancy. Her birth length was 48 cm (60^{th} centile), weight 3140 grams (55^{th} centile), and head circumference 34.5 cm (69^{th} centile). Current head circumference is 48 cm (5^{th} centile), length is 98 cm (5^{th} centile) and weight is 15 kg (17^{th} centile). An EEG was performed because of irritability and sharp waves were noted in left hemispheric region, seizures were observed and treated with valproic acid (150 ml/day). Brain MRI revealed a thin corpus callosum. Gross motor development was delayed and she did not achieve head control until 4 months, did not sit without support until 9-10 months and walked at age of 21 months. At four years of age she only says a few single words.

GPC4 CASE SUMMARY

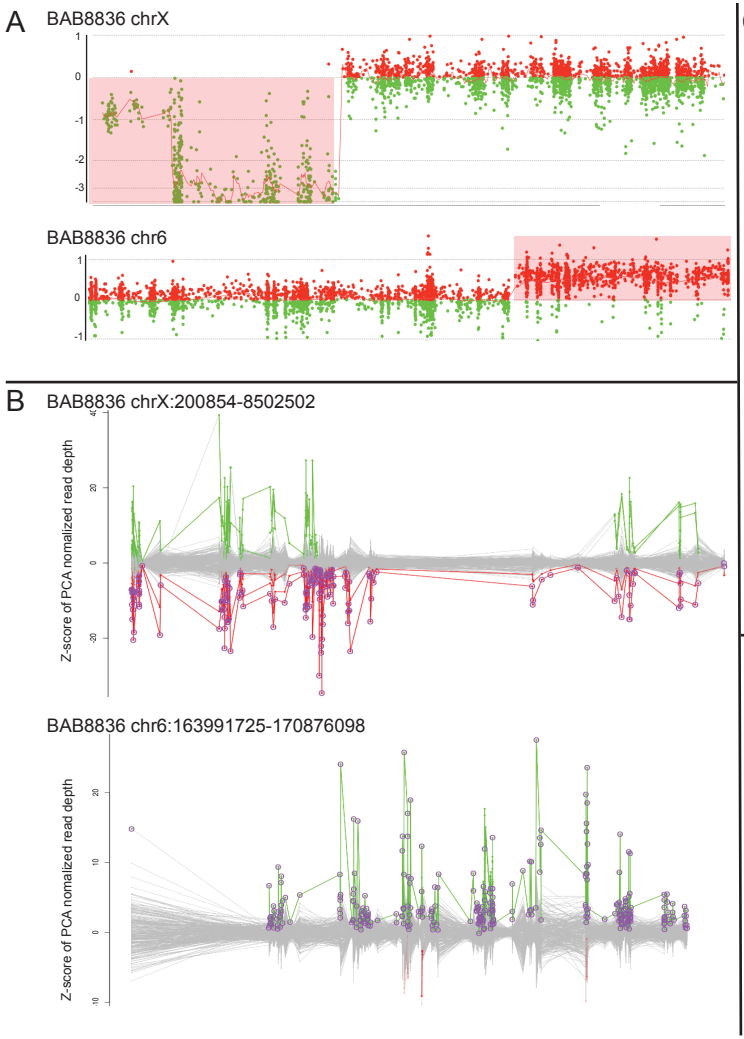
BAB8295

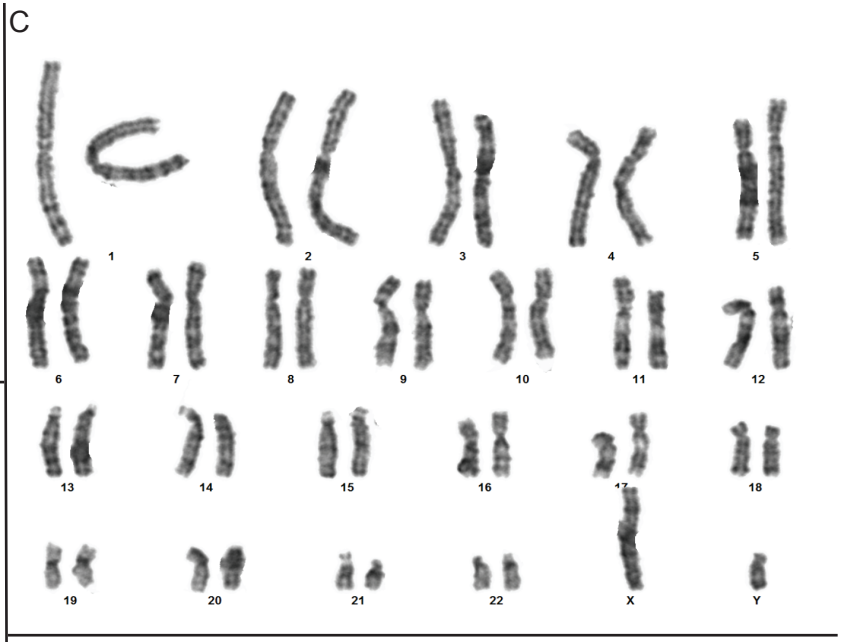
BAB8295 is an 8-year old male. He was a 5-pound-4-ounce (89^{th} centile) product of a 33 week gestation delivered via SVD to 24-year-old G2, P1>2 and 24-year-old father. There were multiple medical issues at birth including choanal atresia, cleft palate, patent ductus arteriosus, grade 2/3 intraventricular hemorrhage, foramen magnum stenosis resulting in palsy of cranial nerves III, VI and VII. Breathing and feeding issues resulted in tracheostomy and gastrostomy tube placement with Nissen fundoplication. He also had recurrent infections managed with IVIG as well as multiple surgeries for the aforementioned birth defects as well as VP shunt placement. Radiographs at that time noted increased skull density and long bone striations. He is presently in 3^{rd} grade in regular classes and makes As. His weight is 23.9 kg (17%), height 115.1 cm (-2.88 SD) and OFC 60.2 cm (+5 SD). He has frontal bossing with a broad forehead, hypertelorism and telecanthus with downslanting palpebral fissures, low nasal bridge, anteverted nares, low-set ears, flat midface, cupid bow mouth, gingival hyperplasia, tracheostomy in place, mesomelia, brachydactyly with spatulated finger tips, broad thumbs and great toes and second toes that are medially deviated. External genitalia were normal. Detailed neuropsychological testing at 7 years of age revealed “average” scores on a variety of standardized tests. DEXA scan at 8 years of age revealed significant cranial sclerosis: Subtotal body (without head) BMC 457.00 g, BMD 0.616 g/cm^2, Z-score -1.0;

head BMC 847.64 g, BMD 2.974 g/cm^2; total body (with head) BMC 1304.64 g, BMD 1.270 g/cm^2, Z-score +9.4.



Supplemental Figure 1
Pedigrees and chromatograms of variants proposed to explain the subjects phenotypes. Yellow highlights the location of identified variants co-segregating in all families with the Robinow features.

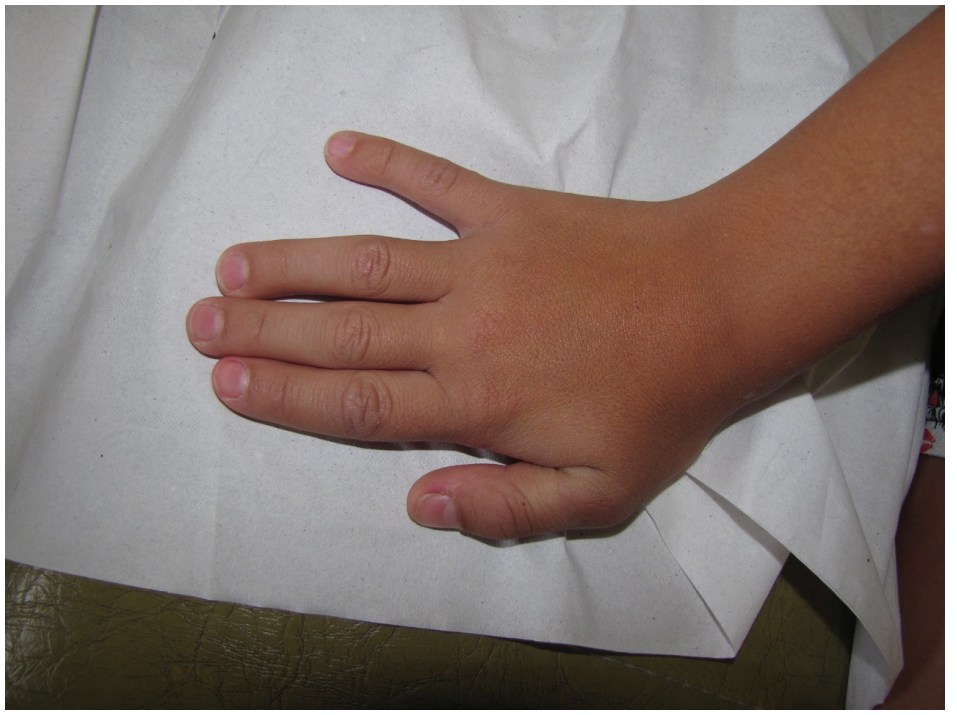


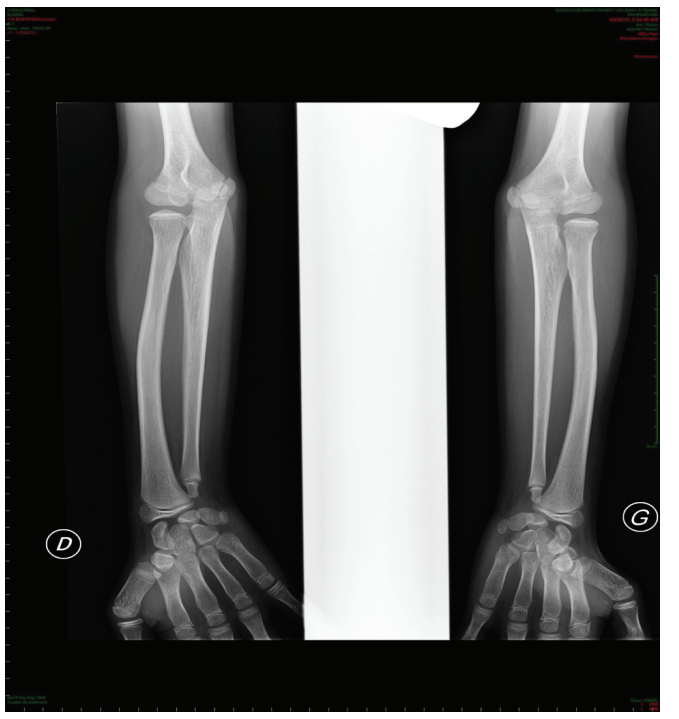


Supplemental Figure 2

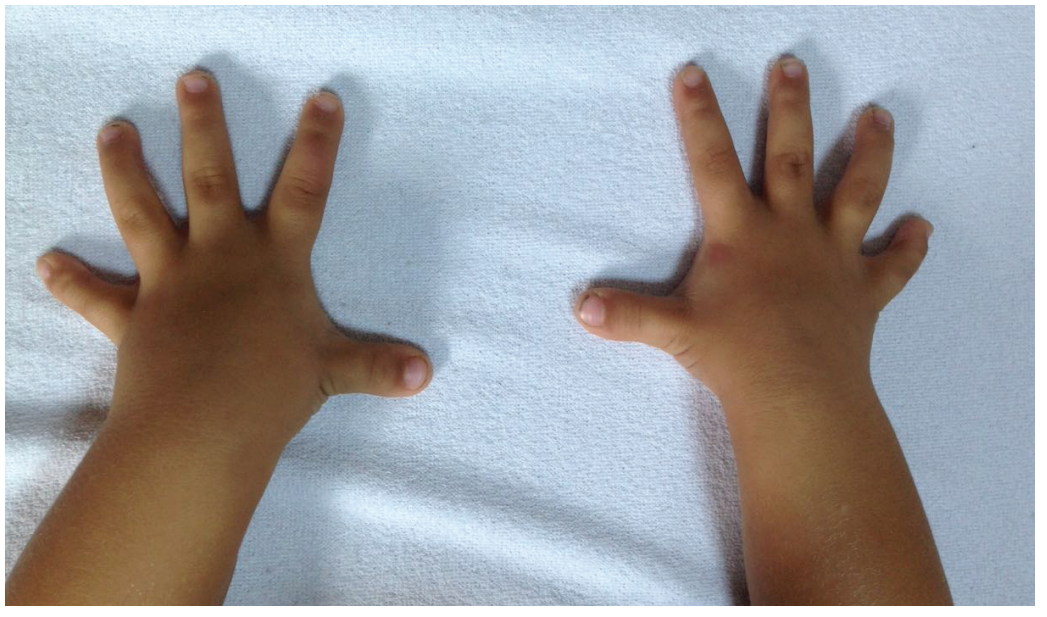
Distinct methodologies used to identify copy-number alterations in BAB8836. A) High-density arrayCGH highlighting a terminal Xp deletion that spans chrX:409876-8199541(min) or chrX:der-8321827(max). A terminal 6q duplication is also observed that spans chr6:157870814-170881475 (min) or chr6:157461178-ter (max). B) XHMM plots identified from exome normalized read-depths showing CNVs in BAB8836. C) Karyotype of BAB8836 with 700 band resolution with a possible derivative X chromosome 46,XY,der(X)t(X,6)(p22.31;q25.3). arr[GRCh37]6q25.3q27(157,870,814_ 170,881,475)x3, Xp22.33p22.31(409,876_8,199,541)x0

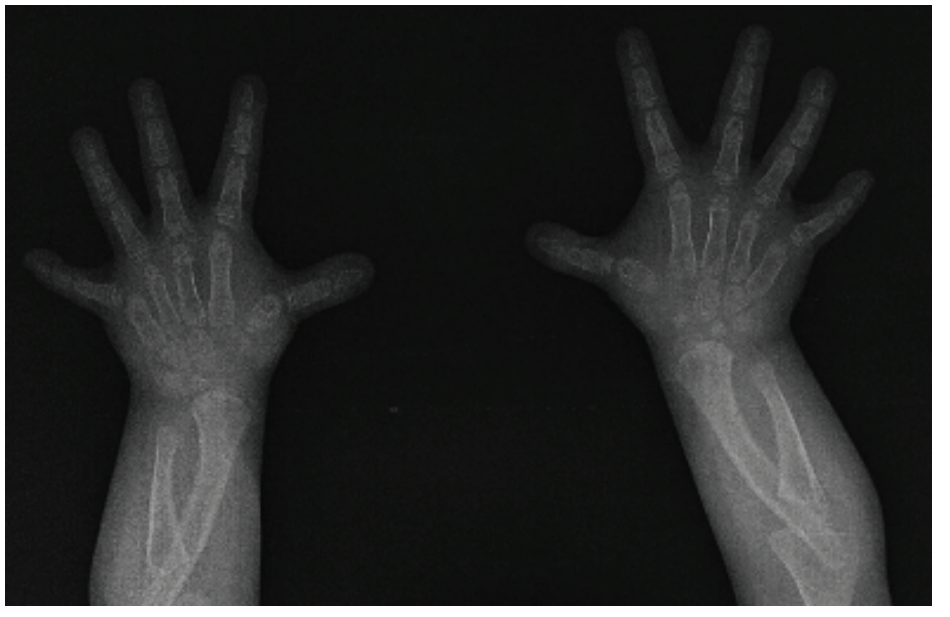
5449



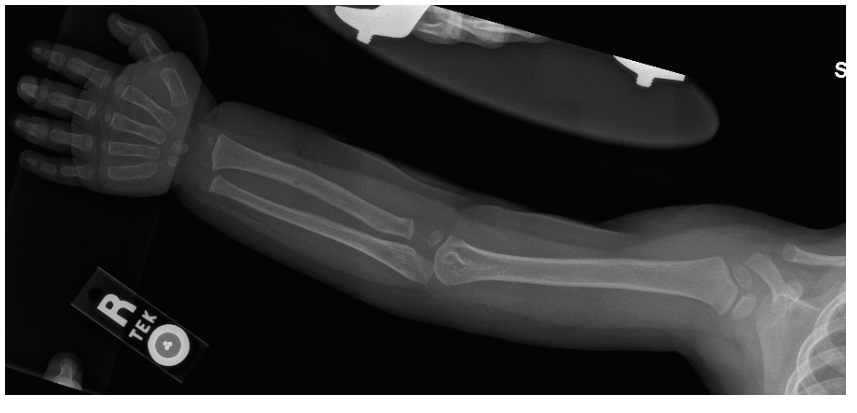


BAB8705

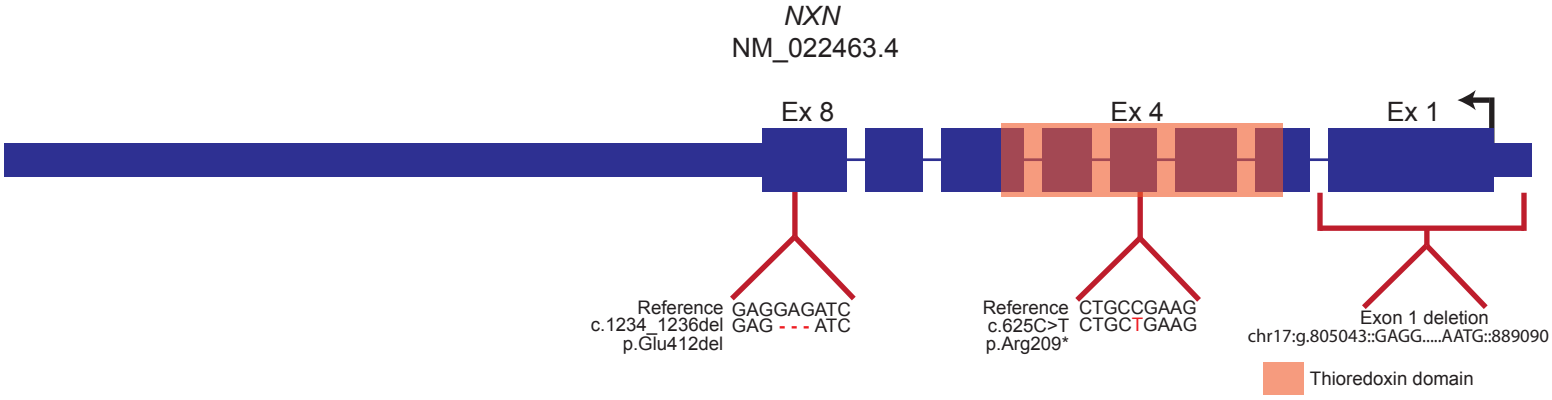




BAB8596



Supplemental Figure 3
Photos and radiographs of available individuals with variants in *FZD2*



Supplemental Figure 4

Locations of the three distinct variants identified in the coding region of *NXN* according to the RefSeq transcript NM_022463.4. Orange box indicates the predicted thioredoxin domain of the resulting protein structure.

Supplemental table 1: Variants reported in this study

Individual ID	Secondary ID	Origin	Relationship	Gene	variant type	Zygosity	Transcript	cds_variant	p_variant	OMIM_phenotype
BAB9126	-	in-house cohort	Affected	*DVL1*	-1 frameshift	Heterozygous	NM_004421.2	c.1612_1616dup	p.Ser539Argfs*112	Robinow syndrome, Autosomal Dominant 2, 616331
BAB9128	-	in-house cohort	Affected	*DVL1*	-1 frameshift	Heterozygous	NM_004421.2	c.1612_1616dup	p.Ser539Argfs*112	Robinow syndrome, Autosomal Dominant 2, 616331
BAB9300	-	in-house cohort	Affected	*DVL1*	-1 frameshift	Heterozygous	NM_004421.2	c.1623del	p.Ser542Valfs*107	Robinow syndrome, Autosomal Dominant 2, 616331
BAB9129	-	in-house cohort	Affected	*DVL1*	-1 frameshift	Heterozygous	NM_004421.2	c.1496_1508del	p.Pro499Argfs*146	Robinow syndrome, Autosomal Dominant 2, 616331
16420	-	in-house cohort	Affected	*DVL1*	-1 frameshift	Heterozygous	NM_004421.2	c.1505_1517del	p.His502Profs*143	Robinow syndrome, Autosomal Dominant 2, 616331
BAB9236	-	in-house cohort	affected	*DVL1*	-1 frameshift	Heterozygous	NM_004421.2	c.1608_1623del	p.Ser537Valfs*107	Robinow syndrome, Autosomal Dominant 2, 616331
BAB9135	-	in-house cohort	Affected	*DVL3*	-1 frameshift	Heterozygous	NM_004423.3	c.1617del	p.Gln539Hisfs*129	Robinow syndrome, Autosomal Dominant 3, 616894
BAB 7987	BH9272_1	in-house cohort	Affected	*FZD2*	missense	Heterozygous	NM_001466.3	c.1301G>T	p.Gly434Val	NA
BAB 7988	BH9272_2	in-house cohort	Affected mother of BAB7987	*FZD2*	missense	Heterozygous	NM_001466.3	c.1301G>T	p.Gly434Val	NA
BAB8594	BH8637_3	in-house cohort	affected mother of BAB8596	*FZD2*	Nonsense	Heterozygous	NM_001466.3	c.1130G>A	p.Trp377*	NA
BAB8596	BH8637_1	in-house cohort	affected	*FZD2*	Nonsense	Compound Heterozygous	NM_001466.3	c.1130G>A c.425C>T	p.Trp377* p.Pro142Leu	NA
BAB8705	BH8808_1	in-house cohort	affected	*FZD2*	missense	Heterozygous	NM_001466.3	c.1301_1302delinsTT	p.Gly434Val	NA
BAB8295	BH7859_1	in-house cohort	affected	*GPC4*	missense	hemizygous	NM_001448.2	c.1235G>A	p.Arg412Lys	NA
BAB9138	BH9278_1	in-house cohort	affected	*WNT5A*	missense	Heterozygous	NM_003392.4	c.479C>G	p.Ser160Cys	Robinow syndrome, Autosomal Dominant 1, 180700
BAB9131	BH9276_1	in-house cohort	Affected	*WNT5A*	nonframeshift insertion	Heterozygous	NM_003392.4	c.487_492dup	p.Gly163_Cys164dup	Robinow syndrome, Autosomal Dominant 1, 180700
BAB8759	BH9282_1	in-house cohort	affected	*INPPL1;SH3PXD2B*	Missense; Frameshift deletion	Homozygous(2)	NM_001567.3, NM_001017995.2	c.1636G>A;c.969del	p.Val546Ile; p.Arg324Glyfs*19	Opsismodysplasia, 258480; Frank-ter Haar syndrome, 249420
BAB8841	BH9279_1	in-house cohort	affected	*NXN*	Nonsense	Homozygous	NM_022463.4	c.625C>T	p.Arg209*	NA
BAB8747	BH8811_1	in-house cohort	affected	*FGD1*	Frameshift insertion	Hemizygous	NM_004463.2	c.892dup	p.Cys298Leufs*5	Aarskog-Scott syndrome, 305400
BAB8751	BH9281_1	in-house cohort	affected	*FGD1*	Frameshift insertion	Hemizygous	NM_004463.2	c.527dup	p.Leu177Thrfs*40	Aarskog-Scott syndrome, 305400
BAB8743	BH8812_1	in-house cohort	affected	*PTPN11*	missense	Heterozygous	NM_002834.4	c.836A>G	p.Tyr279Cys	Noonan syndrome, 163950
BAB8740	BH9280_1	in-house cohort	affected	*RAC3*	missense	Heterozygous	NM_005052.2	c.176C>G	p.Ala59Gly	NA
BAB9127	BH9274_1	in-house cohort	affected	NA	NA	NA		NA		NA
BAB8836	BH10050_1	in-house cohort	affected	NA	CNV	Hemizygous; Heterozygous	NA	46,XY,der(X)t(X;6)(p22.31;q25.3).arr[GRCh37]6q25.3q27(157,870,814_170,881,475)x3, Xp22.33p22.31(409,876_8,199,541)x0	NA	NA
5449	-	GeneMatcher	Affected	*FZD2*	missense	Heterozygous	NM_001466.3	c.1300G>A	p.Gly434Ser	NA
BAB9844	UK1	GeneMatcher	Affected	*NXN*	Nonframeshift deletion, CNV	Compound heterozygous	NM_022463.4	c.1234_1236del; chr17:g.805043::GAGG.....AATG::889090	p.Glu412del,???	NA
BAB9847	UK2	GeneMatcher	Affected	*NXN*	Nonframeshift deletion, CNV	Compound heterozygous	NM_022463.4	c.1234_1236del; chr17:g.805043::GAGG.....AATG::889090	p.Glu412del,???	NA

Supplemental table 2: Variants reported in the literature

Individual ID	Origin	Gene	Transcript	cds_variant	p_variant	OMIM_phenotype
family 1	Roifman et al 2015	*WNT5A*	NM_003392.4	c.257A>G	p.Tyr86Cys	Robinow syndrome, Autosomal Dominant 1, 180700
family 2	Roifman et al 2015	*WNT5A*	NM_003392.4	c.206G>A	p.Cys69Tyr	Robinow syndrome, Autosomal Dominant 1, 180700
family 3	Roifman et al 2015	*WNT5A*	NM_003392.4	c.257A>G	p.Tyr86Cys	Robinow syndrome, Autosomal Dominant 1, 180700
-	Person et al 2010	*WNT5A*	NM_003392.4	c.248G>C	p.Cys83Ser	Robinow syndrome, Autosomal Dominant 1, 180700
-	Person et al 2010	*WNT5A*	NM_003392.4	c.544_545delinsTC	p.Cys182Arg	Robinow syndrome, Autosomal Dominant 1, 180700
-	Saal et al 2015	*FZD2*	NM_001466.3	c.1644G>A	p.Trp548*	NA
BAB4073	White et al 2015	*DVL1*	NM_004421.2	c.1570_1571delins	p.Phe524Profs*125	Robinow syndrome, Autosomal Dominant 2, 616331
BAB4878	White et al 2015	*DVL1*	NM_004421.2	c.1505_1517del	p.His502Profs*143	Robinow syndrome, Autosomal Dominant 2, 616331
BAB5264	White et al 2015	*DVL1*	NM_004421.2	c.1519del	p.Trp507Glyfs*142	Robinow syndrome, Autosomal Dominant 2, 616331
016462	White et al 2015	*DVL1*	NM_004421.2	c.1505_1517del	p.His502Profs*143	Robinow syndrome, Autosomal Dominant 2, 616331
016516	White et al 2015	*DVL1*	NM_004421.2	c.1508del	p.Pro503Argfs*146	Robinow syndrome, Autosomal Dominant 2, 616331
017604	White et al 2015	*DVL1*	NM_004421.2	c.1615del	p.Ser539Alafs*110	Robinow syndrome, Autosomal Dominant 2, 616331
030526	White et al 2015	*DVL1*	NM_004421.2	c.1529del	p.Gly510Valfs*139	Robinow syndrome, Autosomal Dominant 2, 616331
Subject 1	Bunn et al 2015	*DVL1*	NM_004421.2	c.1519del	p.Trp507Glyfs*142	Robinow syndrome, Autosomal Dominant 2, 616331
Subject 2	Bunn et al 2015	*DVL1*	NM_004421.2	c.1562del	p.Pro521Hisfs*128	Robinow syndrome, Autosomal Dominant 2, 616331
Subject 3	Bunn et al 2015	*DVL1*	NM_004421.2	c.1576_1583delinsG	p.Pro526Alafs*121	Robinow syndrome, Autosomal Dominant 2, 616331
BAB8062	White et al 2016	*DVL1*	NM_004421.2	c.1522del	p.Pro508Leufs*141	Robinow syndrome, Autosomal Dominant 2, 616331
015902	White et al 2016	*DVL3*	NM_004423.3	c.1749del	p.Ser583Argfs*85	Robinow syndrome, Autosomal Dominant 3, 616894
BAB7990	White et al 2016	*DVL3*	NM_004423.3	c.1585del	p.Ala529Profs*139	Robinow syndrome, Autosomal Dominant 3, 616894
BAB7985	White et al 2016	*DVL3*	NM_004423.3	c.1715-2A>G	p.?	Robinow syndrome, Autosomal Dominant 3, 616894
BAB7982	White et al 2016	*DVL3*	NM_004423.3	c.1715-1G>A	p.?	Robinow syndrome, Autosomal Dominant 3, 616894
BAB4569	White et al 2016	*DVL3*	NM_004423.3	c.1716del	p.Ser573Valfs*95	Robinow syndrome, Autosomal Dominant 3, 616894