Pathogenic mutations identified by a multimodality approach in 117 Japanese Fanconi anemia patients

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An incorrected version of table 2 appared On October 2019 Issue, page 1967.
The corrected version of table 2 is published on the next page.

Table 2. Clinical phenotype of 10 Japanese Fanconi anemia patients with VACTERL-H association.

Individual	Affected gene	Mutation patterns	VACTERL-H features	FA-features	Family history of FA*	Birth weight SD score	DEB induced chromosome breakage (breaks / cell)	*ALDH2* *** genotype
Case 18-1	*FANCA*	c.2546delC: p.S849FfsX40 c.4042_4043insC: p.I1348TfsX77	C: PDA R: Left renal agenesis L: Bilateral absent thumbs/ Bilateral radial hypoplasia	Short stature	+	-1.9	0.44	AA
Case 30	FANCA	c.2546delC: p.S849FfsX40 c.2546delC: p.S849FfsX40	V: scoliosis C: ASD/Persistent left superior vena E: Esophageal atresia	Skin pigmentation Deafness Right inguinal hernia Bicornuate uterus Short stature (-1.8SD)	–	-2.1	2.06	GG
Case 37	*FANCA*	c.2546delC: p.S849FfsX40 c.3295C>T: p.Q1099X	E: Esophageal atresia R: Right pelvic kidney L: Bilateral thumb hypoplasia	Jejunal atresia Strabismus Short stature (-4SD)	–	-2.3	0.12	GG

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Case 60	*FANCB*	complete loss of *FANCB* gene (chrX g.14730104-14904216 del)	V: Spina bifida occulta/ Abnormal ribs A: Anal atresia C: PDA R: Right renal agenesis L: Right absent thumb/ Partial loss of left thumb	Skin pigmentation Microphthalmus/ Stenocephaly/Ptosis Duodenal stenosis Annular pancreas/ Hypospadias/ Undescended testis Short stature (-6SD)	–	-4.8	3.8	GG
Case 61	*FANCB*	complete loss of *FANCB* gene (chrX g.14810970-14932973 del)	V: Abnormal ribs/Scoliosis A: Anal atresia C: VSD/PS E: Duodenal atresia** R: Left renal agenesis L: Bilateral absent thumbs H: Hydrocephalus	Skin pigmentation Microphthalmus/ Deafness/ Ear canal stenosis Undescended testis (Short stature (-1SD))	–	-2.8	4.2	GA
Case 64	*FANCC*	c.1154+5G>A: p.S386X c.1154+5G>A: p.S386X	A: Anal atresia C: VSD, PDA E: Esophageal atresia	Skin pigmentation Deafness/Left aural stenosis/ Right aural atresia Cleft palate Short stature (-2SD)	–	-2.53	7.8	GG
Case 69	*FANCG*	c.307+1G>C c.1066C>T: p.Q356X	C: Coarctation complex R: Right renal agenesis/ Left renal cyst L: Bilateral absent thumbs/ Right radial hypoplasia	Skin pigmentation Short stature (-8SD)	–	-1.7	8.54	GA
Case 73-1	*FANCG*	c.307+1G>C c.307+1G>C	C: PDA R: Left renal agenesis L: Right absent thumb/ Bilateral radial hypoplasia	Skin pigmentation Bilateral aural atresia Short stature (-2.7SD)	+	-0.9	3.49	GA
Case 96	*FANCI*	c.158-2A>G:p.S54FfsX5 c.288G>A:p.C56FfsX8	A: Anal atresia C: VSD/PDA R: Right renal agenesis/ Left renal hypoplasia L: Bilateral absent thumb/ Bilateral absent radius H: Hydrocephalus	Skin pigmentation Microphthalmus Hypogenitalia Short stature (-8SD)	–	-3.9	0.52	GA
Case 99-1	*FANCP*	c.343delA: p.S115AfsX11 c.343delA: p.S115AfsX11	C: ASD/VSD/PS R: horseshoe kidney L: Bilateral floating thumbs/ bilateral radial hypoplasia	Intestinal malrotation Duodenal stenosis Short stature (-5.8SD)	+	-2.3	0.91	AA

*Case 18-1, 73-1, and 99-1 had a sibling with Fanconi anemia (FA). ** Duodenal atresia is considered to be a part of the VACTERL association by some reports.[27] ***_ALDH2_ wild type and the inactivating mutation (p.Glu504Lys) allele is referred to as G and A, respectively. ALDH2: aldehyde dehydrogenase-2; ASD: atrial septal defect; BM: bone marrow; DEB: diepoxybutane; PDA: patent ductus arteriosus; PS: pulmonary stenosis; SD: Standard Deviation; VACTERL-H: vertebral anomalies, anal atresia, cardiac anomalies, tracheal-esophageal fistula, esophageal atresia, renal structural abnormalities, limb anomalies, and hypocephalus; VSD: ventricular septal defect.

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