

Table S1: Spectrum of congenital abnormalities in 142 patients

ID	Region	Gene	Gender	Diagnosis	Treatment	Consanguinity	Skeletal abnormalities	Facial dysmorphism	Skin changes	Other abnormalities	Relatives with FA
FA-01/ FA-536	Tamil Nadu	FANCG	M	NA	Danazol	Yes	NA	NA	HPOP,Leukoplakia	NA	NA
FA01/19	Kerala	FANCL	M	MDS	BMT	NA	NA	NA	CAL, HP	NA	NA
FA-02	Tamil Nadu	FANCL	M	MDS	Danazol	No	NA	MCH	CAL	NA	NA
FA-03/ FA-515	Assam	FANCA	F	NA	Danazol	NA	TH, TA	DF	HP	NA	NA
FA-03/19	West Bengal	FANCI	M	NA	Stanozolol	No	SS	LSE, MOA, MCH, TF	HP, CAL	NA	Sister
FA-04/ FA-509	Kerala	FANCA	M	NA	Stanozolol	No	BT, Absent thumb	LSE	HP, HPOP, CAL	NA	NA
FA-05/ FA-500	Tamil Nadu	FANCA	M	NA	Stanozolol	Yes	NA	NA	HP	NA	NA
FA-06	West Bengal	FANCA	M	MDS	BMT	NA	SS, TH, Thenar hypoplasia	TF	HPOP, HP	NA	NA
FA-07	Andhra Pradesh	FANCL	M	NA	Stanozolol/BMT	NA	NA	MCH, frontal blossing	HP, HPOP	NA	NA
FA-08/ FA-552	Andhra Pradesh	FANCA	M	DKC	Stanozolol/BMT	Yes	NA	NA	HP, leukoplakia	NA	NA
FA-09/ FA-505	Kerala	FANCA	F	NA	Danazol/BMT	No	NA	NA	HPOP	NA	NA
FA-10	Tamil Nadu	FANCA	F	NA	Danazol	Yes	Clinodactyly	MCH, MGH, HAP	HP, POH	NA	NA
FA-11/ FA-524	Kerala	FANCA	M	NA	Stanozolol	No	SS, Arachinodactyly	DF	HP	NA	NA
FA-12	West Bengal	FANCA	F	NA	Danazol	No	NA	NA	HP, POH	NA	NA
FA-12/19	Kerala	FANCA	M	NA	Stanozolol	No	Hypothenar, clinodactyly, partial syndactyly	HAP	CAL	HSK	NA
FA-13	Kerala	FANCG	M	NA	Stanozolol	NA	NA	MCH,TF	HPOP, HP	HSK	NA
FA-14/ FA-534	Andhra Pradesh	FANCL	M	NA	Stanozolol	No	NA	NA	HP	NA	NA
FA-15 FA-503	Andhra Pradesh	FANCA	F	NA	Danazol/BMT	NA	SS	MCH	HP	ARP	Sister
FA-16	Andhra Pradesh	FANCA	F	NA	Danazol	Yes	SS	MGH	HP	NA	NA
FA-17/19	Tamil Nadu	FANCA	M	NA	Stanozolol	No	NA	MCH	CAL	NA	Brother
FA-18/ FA-551	Andhra Pradesh	FANCA	M	AML	Stanozolol	No	SS	LSE	HP	NA	Sister had AA
FA-18/19	Tamil Nadu	FANCA	M	NA	Stanozolol	No	BT, polydactyly	DF, EF, HAP	HP	NA	Brother
FA18/20	Kerala	FANCL	F	NA	Danazol/BMT	No	Nail dystrophy, Scoliosis	NA	HP, oral leukoplakia, CAL	NA	NA
FA-19	Karnataka	FANCL	M	NA	Stanozolol	NA	BT, SS	MOA, MCH	HP	NA	NA
FA-20	Tamil Nadu	FANCG	M	NA	Stanozolol	Yes	BT, Polydactyly	AE, MGH	HP	NA	NA
FA-20/19	Andhra Pradesh	FANCA	M	NA	Stanozolol	Yes	NA	DF	HP	Ectopic kidney	NA
FA-21/ FA-510	Jharkhand	FANCA	M	NA	Stanozolol	Yes	BT	AE	HP	NA	NA
FA-22	Kerala	FANCA	M	NA	Stanozolol/ BMT	Yes	Polydactyly	DF	Petechiae, POH	NA	NA
FA-23	Tamil Nadu	FANCA	F	NA	NA	No	Thumb Abnormalities	MCH	HPOP, HP	NA	NA
FA-24/ FA-502	Andhra Pradesh	FANCA	M	HCC	Stanozolol/ BMT	No	Thenar hypoplasia	NA	CAL, HP	NA	NA
FA-25	Andhra Pradesh	FANCL	M	NA	Stanozolol/ BMT	No	SS	NA	HP	Ectopic kidney	NA
FA-26/ FA-530	Tamil Nadu	FANCG	M	NA	Stanozolol	Yes	SS, Polydactyly, Clubbing	MCH	HPOP, HP	NA	NA
FA-27	Andhra Pradesh	FANCA	F	NA	Danazol	Yes	NA	NA	HP	NA	NA
FA-27/19	Tamil Nadu	FANCA	M	NA	Stanozolol	Yes	Triphalageal thumb	Hypertelorism, HAP	HP	NA	Sister, 1 death at birth
FA-28	Tamil Nadu	FANCA	F	NA	Danazol	Yes	SS	MCH	HP	NA	NA
FA-29	Chattisgarh	FANCD2	F	NA	BMT	Yes	NA	NA	NA	HSK	NA
FA-30/ FA-501	Tamil Nadu	FANCF	M	NA	Stanozolol	NA	Thenar hypoplasia	LSE, MCH	HPOP, CAL, HP	NA	NA
FA-30/19	Andhra Pradesh	FANCL	F	NA	Danazol	Yes	NA	MCH	HP, HPOP	NA	NA
FA-31/ FA-514	Andhra Pradesh	FANCA	F	NA	Danazol	No	SS	HAP	HP	NA	NA
FA-32	Jharkhand	FANCA	M	NA	Stanozolol	No	SS	LSE	HP, HPOP	NA	NA
FA-33	Tamil Nadu	FANCA	M	NA	Stanozolol/ BMT	Yes	NA	NA	HP	NA	Sister with AA, brother died
FA-34	Kerala	FANCA	M	NA	Stanozolol/ BMT	Yes	SS	TF	CAL, HP	NA	NA
FA-35	Kerala	FANCA	M	NA	BMT	NA	TH, SD	Depressed nasal bridge, MCH	CAL	NA	NA
FA-36/ FA-526	Andhra Pradesh	FANCG	M	NA	Stanozolol	Yes	NA	TF, ASE	CAL, HP	NA	NA
FA-36/19	Tamil Nadu	FANCG	M	AML	Stanozolol	No	TH	HAP, small mouth and eyes, ASE	HP, CAL	Small kidneys	Sister
FA-37/19	Tamil Nadu	FANCG	F	MDS	Danazol	No	NA	HAP, small mouth and eyes, ASE	HP, CAL	NA	Brother
FA-38	Maharashtra	FANCG	F	NA	BMT	No	Subtle thumb abnormality	NA	HP	NA	NA
FA-39 /FA-518	Andhra Pradesh	FANCC	M	NA	Stanozolol	Yes	Thenar hypoplasia	NA	HP	NA	NA
FA-49/19	Andhra Pradesh	FANCL	M	DKC	NA	Yes	SS	NA	HP, HPOP, CAL	Hypoplastic kidneys	NA
FA-511	Andhra Pradesh	FANCA	M	NA	Stanozolol/ BMT	Yes	SST	NA	HP, CAL	NA	Sibling died of FA-AML
FA-513	Andhra Pradesh	FANCA	M	NA	Stanozolol	Yes	TH	LSE	HP, CAL	NA	NA
FA-516	West Bengal	FANCA	M	NA	Stanozolol	Yes	NA	LSE, MCH	CAL	NA	NA
FA-517	Kerala	FANCC	M	NA	Danazol/ BMT	Yes	TA, Ectrodactyly, Clinodactyly	HAP, MOA, MCH	CAL	NA	NA
FA-519	West Bengal	FANCD2	M	NA	Stanozolol	No	SS, Absent simian crease, TH	TF	HP	NA	NA
FA-521	Andhra Pradesh	FANCA	M	NA	Stanozolol	Yes	TH	AE	HP	NA	NA
FA-523	Kerala	FANCL	M	NA	Stanozolol	No	NA	MCH	CAL, HP	NA	NA
FA-525	Tamil Nadu	FANCL	M	NA	Stanozolol/ BMT	No	Syndactyly, DPT	NA	HP, HPOP	NA	NA
FA-527	Chattisgarh	FANCC	F	NA	Danazol/ BMT	No	SS, Clinodactyly	NA	HP	NA	NA
FA-528	Haryana	C17orf70	F	NA	BMT	No	SS	MCH	NA	NA	NA
FA-529	Tamil Nadu	FANCA	F	NA	Danazol	No	SS, BT	MCH	HP	NA	NA

FA-531	Karnataka	FANCG	M	NA	BMT	Yes	Clinodactyly	NA	HP, CAL	ARP	NA
FA-532	Andhra Pradesh	FANCL	F	NA	Danazol	Yes	NA	DF	HP, HPOP	NA	NA
FA-533	Maharashtra	FANCA	F	NA	Danazol/ BMT	No	NA	HAP, MGH	HP	NA	NA
FA-535	Andhra Pradesh	FANCL	M	NA	Stanozolol	Yes	SS	MGH	HP	NA	NA
FA-538	Andhra Pradesh	FANCL	M	NA	Stanozolol	NA	SS, TH	MCH, HAP	CAL	NA	NA
FA-543	Andhra Pradesh	FANCL	F	NA	Danazol	Yes	Nail dystrophy	HAP	HPOP	NA	NA
FA-544	Andhra Pradesh	FANCA	M	NA	Stanozolol	NA	Clinodactyly	LSE, HAP	HPOP, CAL, HP	NA	NA
FA-547	Karnataka	FANCA	F	NA	Danazol	Yes	Skeletal deformity	MOA, DF, MGH	HPOP	NA	NA
FA-548	Kerala	FANCL	M	NA	Stanozolol	No	SS	MCH, DF	HP, HPOP	NA	NA
FA-549	Bihar	FANCA	M	NA	Stanozolol	No	BT, Polydactyly	NA	HP	NA	NA
FA-554	Kerala	FANCA	M	NA	Stanozolol	No	SS, Bilateral clinodactyly	NA	CAL, HP, Pallor	NA	Sister
FA-556	Karnataka	FANCA	M	NA	Stanozolol	No	NA	NA	HP	Kidney abnormalities	NA
FA-557	Kerala	FANCA	M	NA	Stanozolol/ BMT	No	NA	NA	HP, HPOP	NA	NA
FA-561	Tamil Nadu	FANCA	F	MDS	Danazol/ BMT	Yes	SS	Hypertelorism	HP	NA	NA
FA-564	Tamil Nadu	FANCL	M	NA	Stanozolol	Yes	SS	MOA	COP, CAL	NA	NA
FA-565	West Bengal	FANCA	M	MDS	Stanozolol	No	Clinodactyly	MCH, MOA	CAL, Pallor	NA	NA
FA-573	Kerala	FANCA	M	NA	Stanozolol/ Metformin	No	SS	NA	CAL	Hypogonadism	NA
FA-581	Andhra Pradesh	FANCA	M	NA	Stanozolol/ BMT	Yes	SS, TH	MCH, LSE, MOA	POH	NA	NA
FA-588	Kerala	FANCG	M	NA	Stanozolol/ BMT	No	SS, Short neck, thumb abnormalities	MCH, hypertelorism	POH, HP	NA	NA
FA-591	Tamil Nadu	FANCA	F	NA	Danazol	Yes	Thenar and hypothenar atrophy	NA	HP, CAL	NA	yes
FA-592	Andhra Pradesh	FANCA	F	NA	Danazol/ BMT	No	SS	NA	POH, CAL	NA	NA
FA-593	Tamil Nadu	FANCA	M	MDS	Stanozolol	Yes	TH (R), TA (L)	NA	HP	NA	NA
FA-595	Tamil Nadu	FANCL	M	NA	Stanozolol	No	NA	NA	CAL, HPOP	NA	Brother
FA-598	Karnataka	FANCG	M	NA	Stanozolol/ BMT	Yes	SS	NA	CAL	Atrophic left kidney	NA
FA-599	Tamil Nadu	FANCA	F	NA	Danazol	Yes	NA	Small and closely set eyes, HAP	CAL, HP	kidney abnormalities	NA
FA-601	Tamil Nadu	FANCL	M	NA	Danazol/ BMT	No	NA	Oral leukoplakia, MCH	HP, HPOP, POH	NA	Brother
FA-614	Andhra Pradesh	FANCA	M	NA	Stanozolol/ BMT	No	SS, clinodactyly	HAP	HPOP	NA	NA
FA-622	West Bengal	FANCA	F	NA	Danazol	No	NA	NA	HP	NA	NA
FA-629/18	Gujrat	FANCG	M	NA	NA	No	SS, TH	HAP, EF	NA	Absent kidney	NA
FA-631/18	Kerala	FANCA	F	MDS	Danazol	No	NA	DF	NA	NA	Sister died during chemotherapy
FA-636/18	Karnataka	FANCT	F	MDS/AML	NA	No	SS, congenital kyphoscoliosis, SD	MCH, TF	HP	Solitary kidney	NA
FA-637/18	Tamil Nadu	FANCL	M	MDS	BMT	No	NA	NA	NA	Ectopic kidney	NA
FA-638/18	Andhra Pradesh	FANCL	M	NA	Stanozolol	No	Bilateral clinodactyly, SST	LSE, MOA	COP, HP, CAL	NA	NA
FA-641/18	Kerala	FANCF	M	MDS	BMT	No	Hypoplasia of upper and lower limbs	NA	HP, CAL	NA	NA
FA-650/18	Andhra Pradesh	FANCG	F	NA	Danazol	Yes	SS, TA	NA	HP	NA	NA
FA-652/18	West Bengal	FANCA	F	NA	Danazol	Yes	NA	DF	NA	NA	NA
FA-659/18	Tamil Nadu	FANCA	M	NA	Stanozolol/ BMT	Yes	congenital TH, CTEV	MCH, LSE	POH, CAL	Congenital acyanotic heart disease	NA
FA-660/18	Tamil Nadu	FANCA	F	NA	NA (Pregnant)	Yes	Polydactyly	HAP	HP	NA	NA
FA-665/18	Kerala	FANCL	F	NA	Danazol	Yes	SS, clinodactyly	MCH, LSE, HAP	CAL, COP	NA	NA
FA-672/18	Kerala	FANCA	M	NA	Danazol	No	BT	HAP	CAL	NA	NA
FA-674/18	Kerala	FANCA	M	NA	Stanozolol	No	TH	MCH	Patches over body	kidney abnormalities, ARP	NA
FA-675/18	Kerala	FANCL	F	NA	Stanozolol/ BMT	No	NA	MCH, EF	NA	NA	NA
O-117	Tamil Nadu	FANCA	F	NA	Danazol/ BMT	Yes	SS, emaciated	MCH, MOA, LSE, HAP, EF	POH, HPOP	NA	NA
O-123	Kerala	FANCL	F	NA	Danazol	No	BT	MCH	POH, HPOP, CAL	NA	NA
O-126	Andhra Pradesh	FANCA	M	MDS-AML	Stanozolol/ BMT	Yes	Simian crease in both hands	LSE, hypertelorism	HP	NA	NA
FA-40	Tamil Nadu	FANCA	M	NA	NA	Yes	NA	TF	CAL	kidney abnormalities	NA
FA-522	Kerala	FANCL	M	NA	NA	No	Thenar and hypothenar hypoplasia	NA	CAL, HP	NA	NA
FA-541	Tamil Nadu	FANCA	F	NA	BMT	Yes	NA	NA	NA	NA	NA
FA-542	NA	FANCA	F	NA	NA	NA	NA	NA	NA	NA	NA
FA-568	Andhra Pradesh	FANCA	M	MDS	BMT	No	NA	NA	NA	NA	Sibling died
FA-580	Bihar	FANCG	F	NA	Danazol	No	NA	NA	NA	Ectopic kidney	NA
FA-584	Tamil Nadu	FANCA	F	NA	Danazol	Yes	NA	NA	Pigmented tongue	NA	NA
FA-646/18	Andhra Pradesh	FANCA	F	NA	Danazol/ BMT	Yes	NA	NA	NA	Anorectal malformation	Sister died with congenital anomaly
FP-23-P-11	Andhra Pradesh	FANCA	M	NA	Danazol	No	NA	NA	NA	Anorectal malformation	NA
P-177	West Bengal	FANCA	F	NA	Danazol	No	NA	Epicanthus	NA	NA	NA
P-603	Andhra Pradesh	FANCL	M	NA	Stanozolol	Yes	Clinodactyly	LSE, HAP, depressed nasal bridge, MCH	Ichthyosis, CAL, HP	NA	NA
FA-04/20	Kerala	FANCA	F	NA	Stanozolol/ BMT	No	NA	NA	NA	NA	NA

FA-06/20	Telangana	FANCA	F	NA	Danazol	Yes	Polydactyly	HAP, EF	NA	NA	NA
FA-10/20	Andhra Pradesh	FANCA	F	NA	Danazol	Yes	NA	MCH, HAP	HP, CAL	NA	NA
FA-10/21	Andhra Pradesh	FANCA	F	NA	Danazol	Yes	Thenar and hypotherar hypoplasia	MCH, Small and closely set eyes, HAP	CAL	NA	NA
FA-11/20	West Bengal	FANCA	M	NA	Stanozolol	Yes	SS	MCH, TF, HAP	CAL	NA	NA
FA-15/20	Lakshadweep	FANCG	M	NA	Stanozolol	No	SS	NA	HP	ARP	NA
FA-16/20	Andhra Pradesh	FANCA	F	MDS	Danazol	No	NA	NA	NA	NA	NA
FA-21/20	Andhra Pradesh	FANCA	F	MDS	BMT	Yes	NA	MCH	HP	NA	NA
FA-5/21	West Bengal	FANCT	M	MDS	Danazol	No	NA	NA	NA	NA	NA
FA-01/20	Kerala	FANCG	M	NA	Stanozolol	Yes	Clinodactyly	MCH, MGH	NA	Left ectopic kidney	NA
FA-02/21	West Bengal	FANCA	M	NA	Stanozolol	No	SS	NA	NA	NA	NA
FA-08/19	Andhra Pradesh	FANCA	F	MDS	Danazol	Yes	SS, Polydactyly	NA	HP	ongenital acyanotic heart disease	NA
FA-13/19	Kerala	FANCA	F	NA	Danazol	No	Polydactyly, Thumb anomalies	MCH, HAP	CAL	NA	NA
FA-18/21	Tamil Nadu	FANCA	M	MDS	Stanozolol	Yes	SS, TH	EF	HP	NA	NA
FA-21/21	Kerala	FANCA	F	NA	Danazol/BMT	No	NA	NA	HP	NA	NA
FA-23/19	Tamil Nadu	FANCA	M	NA	Stanozolol	Yes	SS	EF, HAP	NA	NA	NA
FA-26/21	Jharkhand	FANCA	M	NA	NA	Yes	Hypotherar wasting	NA	HP	Anorectal malformation	NA
FA-30/21	Tamil Nadu	UBE2T/FANCT	F	NA	NA	Yes	Polydactyly	NA	HP	HSK	NA
FA-31/21	Odisha	FANCA	F	NA	Danazol	NA	NA	EF, HAP	NA	NA	NA
FA-32/19	Kerala	FANCA	M	NA	Stanozolol	Yes	Musculoskeletal abnormalities	Hypertelorism, MCH, HAP	CAL	NA	NA
FA-33/19	Maharashtra	FANCL	M	NA	Danazol	No	NA	NA	HP, CAL	Small testes	NA
FA-33/21	Jharkhand	FANCA	M	NA	NA	Yes	NA	Fanconi facies	CAL	NA	NA
FA-34/19	Kerala	FANCL	M	NA	Stanozolol	No	NA	NA	HP	NA	NA
FA-35/21	Andhra Pradesh	FANCA	F	NA	Danazol	Yes	Thenar hypoplasia	NA	NA	NA	NA
FA-649/18	West Bengal	BRCA2	M	AML	NA	No	Thumb Abnormalities	MCH	HP, HPOP	Wilms tumor	Sibling died of FA-AML
FA-12/22	Andhra Pradesh	FANCL	F	NA	Danazol	Yes	Clinodactyly	TF, MCH	HP	Anorectal malformation	NA

**Skeletal abnormalities:** SS- Short stature; TH- Thumb hypoplasia; BT- Bifid thumb; SST- Short stubby toes; TA- Thumb aplasia; CTEV- Congenital talipes equinovarus; HSK- Horse shoe kidney; DPT- Distally placed thumb; ARP- absent radial pulse; SD- Sprengel's deformity

**Facial Dysmorphism:** DF- Dysmorphic Face; MCH- Microcephaly; MGH- Micrognathia; AE- Almond shaped eyes; Low set ears- LSE; High arched palate- HAP; Arched palate- AP; Elfin Facies- EF; TF- Triangular facies; Microphthalmia- MOA; ASE- Antemongoloid slant of eyes

**Skin changes:** HP- Hyperpigmentation; HPOP- Hypopigmentation; CAL- Café au lait; COP- Circum oral pigmentation; POH- Perioral hypopigmentation

**Other anomalies:** HSK- Horse-shoe kidney