Code		Mutation 1	Mutation 2	Clinical features	Age and type of Cancer	*Outcome*	*Ref*
HSC62	**M**	**IVS19-1 G>A**	**IVS19-1 G>A**	Abnormal thumb (limited clinical description)	None at 30 years	No details reported	***Howlett et al***
EUFA579	**F**	**7235G _ A (p.R2336H)**	**5837TC _ AG (p.F1807X)**	Pigmented, abnormal thumb (limited clinical description*)	AML, 2 years	No details reported	***Howlett et al***
EUFA423	**F**	**7691/insAT [R2488fs]**	**9900/insA**	Pigmented, abnormal thumb, bone marrow failure (limited clinical description)	Brain, 3 years	No details reported	***Howlett et al***
AP37P	**M**	**8415G RT K2729N**	**8732C RA [S2835X]**	Short, pigmented, cafe´ au lait, abnormal thumb, Sprengel deformity, midfacial hypoplasia	AML, 2 years	Died of AML age 3	***Howlett et al Ikdeda et al***
1A	**M**	**6174delT**	**9435T>A [C3069X]**	Short, cafe´-au-lait, pigmented, abnormal thumbs and radii, microcephaly, imperforate anus, epicanthal folds, micropenis, undescended testes, dislocated hips, hydronephrosis, abnormal hearing (VATER*)	Brain tumour medulloblastoma or astrocytoma, 4.9 years	Died of tumor age 5	***Offit et al***
1B	**M**	**6174delT**	**9435T>A [C3069X]**	Short, abnormal thumbs, microcephaly, imperforate anus with rectovaginal fistula, slanted eyes, anomalous kidneys, small ear, hip dysplasia (VATER*)	Astrocytoma 2 years,	Died of tumour age 2	***Offit et al***
2	**F**	**6174delT**	**886delG**	No detailed clinical features reported	Medulloblastoma, 4.5 years	No details reported	***Offit et al***
3	**F**	**5301insA**	**7690T RC [I2490T**	No detailed clinical features reported	Medulloblastoma 2.5 years	No details reported	***Offit et al***
4	**F**	**4150G>T [E1308X]**	**9424C>T [Q3066X]**	No detailed clinical features reported	Medulloblastoma, 3.5 years	No details reported	***Offit et al***
K1S1	**M**	**886delGT**	**8447T RA [L2740X]**	Cafe´ au lait, microcephaly, Cardiac malformation	Medulloblastoma, 2.3 years	Died of treatment toxicity	***Hirsch et al***

K1S2	**M**	**886delGT**	**8447T RA [L2740X]**	Cafe´ au lait, abnormal facies, epicanthus (limited clinical description)	M Wilms' tumour, 1.3 years Medulloblastoma, 4.3 years	Died of progressive medulloblastoma	***Hirsch et al***
K2S1	**M**	**4876G RT [E1550X]**	**7757T RC [L2510P]**	Short, microcephaly (limited clinical description)	M Wilms' Tumour, 0.5 years, AML, 2 years	Died of refractory AML and toxicity age 2	***Hirsch et al***
K2S2	**F**	**4876G RT [E1550X]**	**7757T RC [L2510P]**	Short, pigmented, bifid thumb, elfin facies, small palpebral fissures	T-ALL, 4.9 years	remission from T-Cell leukaemia age 5, no further outcome reported	***Hirsch et al***
129/1	**Not reported**	**IVS7+2T>G**	**IVS7+2T>G**	Short, IUGR, cafe´ au lait, microcephaly, imperforate anus	AML, 2.2 years	Died of refractory AML	***Wagner et al***
357/1 A	**Not reported**	**8106G RC [W2626C]**	**2041insA**	Short, hypoplastic thumb, imperforate anus	AML, 1.9 years	Died of refractory AML	***Wagner et al***
632/1	**F**	**IVS7+1G>A**	**5910C RG [Y1894X]**	Short, cafe´ au lait, dysplastic hips, pelvic kidney	AML, 3 years	Died of refractory AML	***Wagner et al***
632/2	**F**	**IVS7+1G>A**	**5910C RG [Y1894X]**	Short, imperforate anus, hypoplastic thumb	AML, 21 months	Refractory AML, alive 6 at 6 months follow up. No further outcome reported.	***Wagner et al***
800/1	**M**	**IVS7+2T >G**	**5164del4**	IUGR, microcephaly, FTT, micropenis, cafe´ au lait spots	AML, 0.9 years	Died of refractory AML	***Wagner et al***
800/2	**M**	**IVS7+2T >G**	**5164del4**	IUGR, microcephaly, cafe´ au lait spots, FTT	Wilms' Tumour 0.8 years	Alive 9 months follow up. No further FU reported	***Wagner et al***
RB		**886delTG**	**5873C RA [S1882X]**	Short, pigmented, cafe´ au lait spots, microcephaly, cryptorchidism	Wilms' Tumour 3.5 years, glioblastoma	Died age 10 of brain tumour	***Reid et al***

					multiforme, 9 years		
CB		**886DelTG**	**5873C RA** **[S1882X]**	cafe´ au lait spots (limited clinical description)	Wilms' (0.6), Brain— medulloblastoma (6), B-ALL (10)	Died age 12 of progressive medulloblastoma	***Reid et al***
SB1690CB		**IVS7+2T>G**	**3827delGT**	Hypermobile thumb, microcephaly, imperforate anus, deafness, renal dysplasia, midfacial hypoplasia (VATER*)	AML 2.1 years	Died of progressive AML age 2	***Meyer et al***
NCI 1		**6174delT**	**9424C RT** **[Q3066X]**	Short, cafe´ au lait, microcephaly, facial dysmorphism, abnormal thumbs, anterior anus, cloudy corneas, ectopic kidneys, delayed development, hydrocephaly (VATER*)	Medulloblastoma, 3.1 years	Not reported	***Alter et al***
P5		**c.2860 A>T**	**c.7964 A>G**	Dislocated hips, facial abnormalities, café au lait spots, growth retardation	Medulloblastoma, 3.5 years	Died of progressive medulloblastoma age 5	***Bodd et al***
PT2		**1548del4**	**1548del4**	Short, pigmented, cafe´ au lait,spots, adducted thumbs, microcephaly, sacral hemivertebra, ventricular septal defect, pelvic kidney, oesophageal atresia, micrognathia, CNS gyrations, congenital cataract (VATER*)	Nephroblastoma, Bilat. Neuroblastoma, Posterior fossa tumour before 1.5 years	Died age 16 months	***Faivre et al***
1703		**c.7878G>C (p.W2626C)**	**c.9097dupA p.T3033NfsX10**	vertebral, anal, cardiac, tracheal, renal and limb anomalies with hydrocephalus. VACTERL-H	Hepatoblastoma, 4 years	Died of treatment complications age 4	***Kopic et al***
Pt 1	**11**	**g.3492insT (c.3264dupT)**	**g.8715þ3A>G (c.8487þ3A>G)**	Microcephaly; esotropia; cerebellar hypoplasia; arachnoid cyst; abnormal radii and thumbs; clinodactyly; FTT; cafe´ au lait spots	Bilateral Wilms' Tumour , 1 year myelodysplasia 2 years, , medulloblastoma 2 years	*Died of progressive medulloblastoma age 3*	***Myers et al***
Pt 2	**15**	**g.2041insA (c.1813dupA),**	**g.859þ2A>G (c.631þ2T>G**	Intestinal duplication cysts/mesenteric lymphangioma jejunum; FTT; microcephaly; cafe´ au lait spots; ear anomalies; bilateral clinodactly	Neuroblastoma stage IIb , 17 months, AML 20 months	*Died of refractory AML*	***Myers et al***
Pt 3	**2**	**g.3127delCT**	**g.7235G>C**	Holoprosencephaly; microcephaly; TEF; FTT; ear anomalies;	Differentiating	*Died of refractory*	***Myers et al***

		(c.2899-2900delCT)	**(c.7007G>A)**	sensineural hearing loss; pelvic kidneys; optic anomalies; polydactyly; cafe´ au lait spots; congenital nevus	neuronal neoplasm 21 months , MDS/ AML 24 months	*AML*	
ID12 S1		**6174delT**	**886delGT**	No features reported	Medulloblastoma, 21 months	*Alive 23 moths from diagnosis, no further follow up reported*	***DeWire et al***
ID12 S2		**6174delT**	**886delGT**	No features reported	Medulloblastoma, 15 moths	*Died*	***DeWire et al***
ID 13		**3492insT**	**IVS19+3A>G**	No features reported	Medulloblastoma, 24 months	*Died*	***DeWire et al***