

Supplementary table 2. The mutation in 43 genes in the 40 patients.

Gene	Transcript	cDNA	Amino acid
ATRX	NM_000489	c.C6614T	p.T2205I
	NM_000489	c.3G>A	p.M1
	NM_000489	c.748C>T:	p.Arg250Ter
	NM_000489	c.4810-1G>C	
	NM_138270	c.C6500T	p.T2167I
	NM_000489.3:	c.958delA	p.Thr320LeufsTer12
	NM_138270	c.G7116T	p.M2372I
	NM_000489	c.G7230T	p.M2410I
	NM_000489.3:	c.4829C>T	p.Thr1610Ile
	NM_000489.3	c.1888G>T	p.Gly630Ter
NM_000489.3	c.4746_4747insT	p.Lys1583Ter	
NM_000489.3	c.666dupG	p.Cys223ValfsTer11	
NM_000489.3	c.6193_6194insG	p.Lys2065ArgfsTer3	
DAXX	NM_001141970.1	:c.1948C>T	p.Gln650Ter
	NM_001141970.1	c.1375dupG	p.Glu459GlyfsTer16
	NM_001141970.1	c.1888delT	p.Ser630LeufsTer27
	NM_001141970.1	c.2154_2155insA	p.Ser719IlefsTer34
	NM_001141970.1	c.958C>T	p.Gln320Ter
	NM_001141970.1	c.328_329delCT	p.Leu110ValfsTer13
	NM_001141970.1	c.1701dupG	p.Thr568AspfsTer13;
	NM_000244	c.G527A	p.R176Q
MEN1	NM_000244	c.1313delA	p.H438fs
	NM_000244	c.T38A	p.L13Q
	NM_000244	c.1225delC	p.L409fs
	NM_000244	c.1680_1683del	p.S560fs
	NM_000244	c.C866T	p.A289V
	NM_000244	c.C781T	p.L261F

	NM_000244	c.C978A	p.Y326X
	NM_000244	c.G118A	p.V40M
	NM_000244	c.1561dupC	p.R521fs
	NM_000244.3	c.86G>A	
	NM_130799	c.G512A	p.R171Q
	NM_130799	c.1298delA	p.H433fs
	NM_130799	c.T38A	p.L13Q
	NM_130799	c.1210delC	p.L404fs
	NM_130799	c.1665_1668del	p.S555fs
	NM_130799	c.C851T	p.A284V
	NM_130799	c.C766T	p.L256F
	NM_130799	c.C963A	p.Y321X
	NM_130799	c.G118A	p.V40M
	NM_130799	c.1546dupC	p.R516fs
	NM_130800	c.G527A	p.R176Q,
	NM_130800	c.1313delA	p.H438fs
	NM_130800	c.T38A	p.L13Q
	NM_130800	c.1225delC	p.L409fs
	NM_130800	c.1680_1683del	p.S560fs
	NM_130800	c.C866T	p.A289V
	NM_130800	c.C781T	p.L261F
	NM_130800	c.C978A	p.Y326X
	NM_130800	c.G118A	p.V40M
	NM_130800	c.1561dupC	p.R521fs
	NM_130801	c.G527A	p.R176Q
	NM_130801	c.1313delA	p.H438fs
	NM_130801	c.T38A	p.L13Q
	NM_130801	c.1225delC	p.L409fs
	NM_130801	c.1680_1683del	p.S560fs
	NM_130801	c.C866T	p.A289V
	NM_130801	c.C781T	p.L261F
	NM_130801	c.C978A	p.Y326X
	NM_130801	c.G118A	p.V40M
	NM_130801	c.1561dupC	p.R521fs
	NM_130802	c.G527A	p.R176Q
	NM_130802	c.1313delA	p.H438fs
	NM_130802	c.T38A	p.L13Q

	NM_130802	c.1225delC	p.L409fs
	NM_130802	c.1680_1683del	p.S560fs
	NM_130802	c.C866T	p.A289V
	NM_130802	c.C781T	p.L261F
	NM_130802	c.C978A	p.Y326X
	NM_130802	c.G118A	p.V40M
	NM_130802	c.1561dupC	p.R521fs
	NM_130803	c.G527A	p.R176Q
	NM_130803	c.1313delA	p.H438fs
	NM_130803	c.T38A	p.L13Q
	NM_130803	c.1225delC	p.L409fs
	NM_130803	c.1680_1683del	p.S560fs
	NM_130803	c.C866T	p.A289V
	NM_130803	c.C781T	p.L261F
	NM_130803	c.C978A	p.Y326X
	NM_130803	c.G118A	p.V40M
	NM_130803	c.1561dupC	p.R521fs
	NM_130804	c.1313delA	p.H438fs
	NM_130804	c.G527A	p.R176Q
	NM_130804	c.T38A	p.L13Q
	NM_130804	c.1225delC	p.L409fs
	NM_130804	c.1680_1683del	p.S560fs
	NM_130804	c.C866T	p.A289V
	NM_130804	c.C781T	p.L261F
	NM_130804	c.C978A	p.Y326X
	NM_130804	c.G118A	p.V40M
	NM_130804	c.1561dupC	p.R521fs
	NP_000235.2		p.Arg29Gln
PIK3CA	NM_006218	c.813+1G>A	
	NM_006218	c.G2846A	p.R949Q
	NM_006218	c.G1664A	p.R555K
PTEN	NM_000314	c.T528G	p.Y176X
	NM_000314	c.G821A	p.W274X
	NM_000314	c.317delA	
	NP_000305.3		p.Asp107IlefsTer6
TSC1	NM_001162426	c.G2734A	p.E912K
	NM_001162426	c.G2656A	p.E886K

	NM_001162427	c.G2584A	p.E862K
	NM_001162427	c.G2506A	p.E836K
	NM_000368	c.G2737A	p.E913K
	NM_000368	c.G2659A	p.E887K
TSC2	NM_001077183	c.G3956T	p.S1319I
	NM_001077183	c.5026_5043del	p.1676_1681del
	NM_001077183	c.A4751G	p.N1584S
	NM_001077183	c.C2303T	p.P768L
	NM_001077183	c.C3890T	p.S1297F
	NM_001077183	c.C2962T	p.R988X
	NM_001077183	c.G1373A	p.R458Q
	NM_001077183	c.C4150T	p.R1384C
	NM_001114382	c.G4088T	p.S1363I
	NM_001114382	c.5158_5175del	p.1720_1725del
	NM_001114382	c.A4883G	p.N1628S
	NM_001114382	c.C2303T	p.P768L
	NM_001114382	c.C4022T	p.S1341F
	NM_001114382	c.C3094T	p.R1032X
	NM_001114382	c.G1373A	p.R458Q
	NM_001114382	c.C4282T	p.R1428C
	NM_000548	c.G4157T	p.S1386I
	NM_000548	c.5227_5244del	p.1743_1748del
	NM_000548	c.A4952G	p.N1651S
	NM_000548	c.C2303T	p.P768L
	NM_000548	c.C4091T	p.S1364F
	NM_000548	c.C3094T	p.R1032X
	NM_000548	c.G1373A	p.R458Q
	NM_000548	c.C4351T	p.R1451C
VHL	NM_000551.3	c.341-6C>T	
	NM_000551.3	c.580G>A	
	NM_000551.3	c.431G>A	
	NM_000551.3	c.451_452insC	
	NM_000551.3	c.340+7G>A	
	NM_000551.3	c.340+4C>T	
	NM_000551.3	c.574C>T	
	NP_000542.1		p.Val194Met
	NP_000542.1		p.Gly144Glu

	NP_000542.1		p.Ile151ThrfsTer23
	NP_000542.1		p.Pro192Ser
AKT1	NM_001014431	c.G469A	p.G157S
	NM_001014431	c.C1292T	p.S431L
	NM_001014431	c.C1355T	p.P452L
	NM_001014432	c.G469A	p.G157S
	NM_001014432	c.C1292T	p.S431L
	NM_001014432	c.C1355T	p.P452L
	NM_005163	c.G469A	p.G157S
	NM_005163	c.C1292T	p.S431L
	NM_005163	c.C1355T	p.P452L
ALCAM	NM_001243280	c.A1466G	p.N489S
	NM_001243281	c.A1466G	p.N489S
	NM_001627	c.A1466G	p.N489S
ANGPT1	NM_001146	c.C346T	p.Q116X
	NM_001199859	c.C346T	p.Q116X
ANGPT2	NM_001118887	c.C274T	p.Q92X
	NM_001118887	c.700dupA	p.I234fs
	NM_001118887	c.G1059T	p.W353C
	NM_001118887	c.C272T	p.T91I
	NM_001118887	c.A940G	p.M314V
	NM_001118887	c.700dupA	p.I234fs
	NM_001118888	c.C274T	p.Q92X
	NM_001118888	c.544dupA	p.I182fs
	NM_001118888	c.G906T	p.W302C
	NM_001118888	c.C272T	p.T91I
	NM_001118888	c.A787G	p.M263V
	NM_001118888	c.544dupA	p.I182fs
	NM_001147	c.C274T	p.Q92X
	NM_001147	c.700dupA	p.I234fs;
	NM_001147	c.G1062T	p.W354C
	NM_001147	c.C272T	p.T91I
	NM_001147	c.A943G	p.M315V
	NM_001147	c.700dupA	p.I234fs
ARID1A	NM_006015	c.3977_3978insGCA	p.P1326delinsPQ
	NM_006015	c.G2390A	p.G797D
	NM_006015	c.C4972T	p.R1658W

	NM_006015	c.C6052T	p.H2018Y
	NM_006015	c.C6617T	p.T2206I
	NM_006015	c.C1528T	p.Q510X
	NM_006015	c.C1534T	p.Q512X
	NM_006015	c.T6148G	p.W2050G
	NM_006015	c.C2087T	p.S696F
	NM_006015	c.T3499G	p.S1167A
	NM_006015	c.C5011T	p.R1671W
	NM_006015	c.C1474T	p.Q492X
	NM_006015	c.G4656T	p.Q1552H
	NM_139135	c.3977_3978insGCA	p.P1326delinsPQ
	NM_139135	c.G2390A	p.G797D;
	NM_139135	c.C4321T	p.R1441W
	NM_139135	c.C5401T	p.H1801Y
	NM_139135	c.C5966T	p.T1989I
	NM_139135	c.C1528T	p.Q510X;
	NM_139135	c.C1534T	p.Q512X
	NM_139135	c.T5497G	p.W1833G
	NM_139135	c.C2087T	p.S696F
	NM_139135	c.T3499G	p.S1167A
	NM_139135	c.C4360T	p.R1454W
	NM_139135	c.C1474T	p.Q492X
ASCL1	NM_004316	c.149_150insGCA	p.A50delinsAQ
	NM_004316	c.149_150insGCAGCAGCA	p.A50delinsAQQQ
	NM_004316	c.149_150insGCAGCAGCA	p.A51delinsAQQQ
	NM_004316	c.149_150insGCAGCAGCA	p.A52delinsAQQQ
	NM_004316	c.C55T	p.Q19X
ATM	NM_000051	c.C1537T	p.Q513X
	NM_000051	c.C3883T	p.L1295F
	NM_000051	c.C2149T	p.R717W
	NM_000051	c.T3074C	p.F1025S
CCND1	NM_053056	c.A856C	p.T286P
CCND3	NM_001136017	c.G257T	p.R86L
	NM_001136125	c.G284T	p.R95L
	NM_001287427	c.G350T	p.R117L
	NM_001760	c.G500T	p.R167L
CCNE2	NM_057749	c.165+1G>A	

CDKN1B	NM_004064	c.G228A	p.W76X
	NM_004064	c.G40A	p.E14K
FOXA2	NM_021784	c.C1177T	p.Q393X
	NM_153675	c.C1159T	p.Q387
	NM_021784	c.C515T	p.S172L
	NM_153675	c.C497T	p.S166L
HIF1A	NM_001243084	c.C2221T	p.Q741X
	NM_001530	c.C2149T	p.Q717X
	NM_181054	c.C2149T	p.Q717X
	NM_001243084	c.G1369A	p.D457N
	NM_001530	c.G1297A	p.D433N
	NM_181054	c.G1297A	p.D433N
	NM_001243084	c.C2254T	p.L752F
	NM_001530	c.C2182T	p.L728F
	NM_181054	c.C2182T	p.L728F
KIT	NM_000222	c.G1861A	p.A621T
	NM_001093772	c.G1849A	p.A617T
	NM_000222	c.G1793A	p.G598E
	NM_000222	c.C2318T	p.S773F
	NM_001093772	c.G1781A	p.G594E
	NM_001093772	c.C2306T	p.S769F
MGMT	NM_002412	c.G611A	p.G204D
	NM_002412	c.C475T	p.R159X
MTOR	NM_004958	c.5490_5501del	p.1830_1834del
	NM_004958	c.5490_5501del	p.1830_1834del
	NM_004958	c.C3949T	p.L1317F
	NM_004958	c.G6671A	p.R2224K
	NM_004958	c.G6947A	p.R2316Q
	NM_004958	c.G1667A	p.G556D
	NM_004958	c.A2918C	p.Q973P
	NM_004958	c.C3721T	p.Q1241X
	NM_004958	c.C3458T	p.S1153F
NF1	NM_000267	c.C1726T	p.Q576X
	NM_001042492	c.C1726T	p.Q576X
	NM_000267	c.3668delT	p.I1223fs
	NM_001042492	c.3668delT	p.I1223fs
	NM_000267	c.C8098T	p.Q2700X

	NM_001042492	c.C8161T	p.Q2721X
	NM_000267	c.C5639T	p.P1880L
	NM_001042492	c.C5702T	p.P1901L
	NM_000267	c.C715T	p.Q239X
	NM_001042492	c.C715T	p.Q239X
	NM_001128147	c.C715T	p.Q239X
	NM_000267	c.G1549A	p.E517K
	NM_001042492	c.G1549A	p.E517K
	NM_001128147	c.G1549A	p.E517K
	NM_001042492	c.2002-1G>A	
	NM_000267	c.2002-1G>A	
	NM_001042492	c.7189+1G>A	
	NM_000267	c.7126+1G>A	
NF2	NM_181830	c.G184A	p.A62T
	NM_181831	c.G184A	p.A62T
	NM_181828	c.G307A	p.A103T
	NM_181829	c.G310A	p.A104T
	NM_000268	c.G433A	p.A145T
	NM_016418	c.G433A	p.A145T
	NM_181825	c.G433A	p.A145T
	NM_181832	c.G433A	p.A145T
	NM_181833	c.G433A	p.A145T
	NM_181830	c.C242T	p.A81V
	NM_181831	c.C242T	p.A81V
	NM_181828	c.C365T	p.A122V
	NM_181829	c.C368T	p.A123V
	NM_000268	c.C491T	p.A164V
	NM_016418	c.C491T	p.A164V
	NM_181825	c.C491T	p.A164V
	NM_181832	c.C491T	p.A164V
	NM_181830	c.G1426A	p.D476N
	NM_181831	c.G1426A	p.D476N
	NM_181828	c.G1549A	p.D517N
	NM_181829	c.G1552A	p.D518N
	NM_000268	c.G1675A	p.D559N
	NM_016418	c.G1675A	p.D559N
	NM_181825	c.G1675A	p.D559N

	NM_181832	c.G1675A	p.D559N
PDGFRA	NM_006206	c.2440-1G>A	
PSIP1	NM_021144	c.753delA	p.K251fs
	NM_001128217	c.753delA	p.K251fs
	NM_033222	c.753delA	p.K251fs
	NM_001128217	c.394-1G>A	
	NM_033222	c.394-1G>A	
	NM_021144	c.394-1G>A	
	NM_001128217	c.G1118A	p.C373Y
	NM_033222	c.G1118A	p.C373Y
	NM_001128217	c.858+1G>A	
	NM_033222	c.858+1G>A	
	NM_021144	c.858+1G>A	
RB1	NM_000321	c.G2236A	p.E746K
	NM_000321	c.G2224A	p.V742I
	NM_000321	c.C481T	p.L161F
	NM_000321	c.G1421A	p.S474N
RET	NM_020630	c.C3115T	p.P1039S
	NM_020975	c.C3115T	p.P1039S
TEK	NM_001290078	c.C2563T	p.R855C
	NM_001290077	c.C2878T	p.R960C
	NM_000459	c.C3007T	p.R1003C
	NM_001290078	c.T836A	p.V279E
	NM_001290077	c.T1148A	p.V383E
	NM_000459	c.T1277A	p.V426E
	NM_001290078	c.G1108A	p.V370M
	NM_001290077	c.G1420A	p.V474M
	NM_000459	c.G1549A	p.V517M
THBS1	NM_003246	c.1026+1G>A	
	NM_003246	c.G2774A	p.G925D
	NM_003246	c.G3230A	p.R1077Q
	NM_003246	c.C3164T	p.A1055V
TP53	NM_000546.5	c.1135_1136insA	
	NP_000537.3		p.Arg379GlnfsTer3
	NM_001126115	c.G347A	p.R116Q
	NM_001126116	c.G347A	p.R116Q
	NM_001126117	c.G347A	p.R116Q

	NM_001276697	c.G266A	p.R89Q
	NM_001276698	c.G266A	p.R89Q
	NM_001276699	c.G266A	p.R89Q
	NM_001126118	c.G626A	p.R209Q
	NM_000546	c.G743A	p.R248Q
	NM_001126112	c.G743A	p.R248Q
	NM_001126113	c.G743A	p.R248Q
	NM_001126114	c.G743A	p.R248Q
	NM_001276695	c.G626A	p.R209Q
	NM_001276696	c.G626A	p.R209Q
	NM_001276760	c.G626A	p.R209Q
	NM_001276761	c.G626A	p.R209Q
	NM_000546.5	c.689C>T	
	NP_000537.3		p.Thr230Ile
	NM_000546.5	c.391A>T	
	NP_000537.3		p.Asn131Tyr
	NM_000546.5	c.916C>T	
	NP_000537.3		p.Arg306Ter
	NM_000546.5	c.309C>G	
	NP_000537.3		p.Tyr103Ter
	NM_001126117	c.C613T	p.R205X
	NM_001276699	c.C532T	p.R178X
	NM_001126113	c.C1009T	p.R337X
	NM_001276695	c.C892T	p.R298X
	NM_001126115	c.C241T	p.R81X
	NM_001126116	c.C241T	p.R81X
	NM_001126117	c.C241T	p.R81X
	NM_001276697	c.C160T	p.R54X
	NM_001276698	c.C160T	p.R54X
	NM_001276699	c.C160T	p.R54X
	NM_001126118	c.C520T	p.R174X
	NM_000546	c.C637T	p.R213X
	NM_001126112	c.C637T	p.R213X
	NM_001126113	c.C637T	p.R213X
	NM_001126114	c.C637T	p.R213X
	NM_001276695	c.C520T	p.R174X
	NM_001276696	c.C520T	p.R174X

	NM_001276760	c.C520T	p.R174X
	NM_001276761	c.C520T	p.R174X