

This test contains variants amounts 2038, as described in the following table:

<b>Disease: Thanatophoric dysplasia, type I</b>	<b>Gene: FGFR3</b>	Gene OMIM Number: 134934	NM ID: NM_000142.4	NO. of Variants:5	Phenotype OMIM Number: 187600
Variant: c.1948A>G, c.742C>T, c.1108G>T, c.1118A>G, c.746C>G					
<b>Disease: Thanatophoric dysplasia, type II</b>	<b>Gene: FGFR3</b>	Gene OMIM Number: 134934	NM ID: NM_000142.4	NO. of Variants:1	Phenotype OMIM Number: 187601
Variant: c.1948A>G					
<b>Disease: Cardiofaciocutaneous syndrome IV</b>	<b>Gene: MAP2K2</b>	Gene OMIM Number: 601263	NM ID: NM_030662.3	NO. of Variants:4	Phenotype OMIM Number: 615280
Variant: c.401A>G, c.383C>A, c.170T>G, c.169T>G					
<b>Disease: Cardiofaciocutaneous syndrome III</b>	<b>Gene: MAP2K1</b>	Gene OMIM Number: 176872	NM ID: NM_002755.3	NO. of Variants:4	Phenotype OMIM Number: 615279
Variant: c.158T>C, c.199G>A, c.607G>C, c.389A>G					
<b>Disease: Cardiofaciocutaneous syndrome II</b>	<b>Gene: KRAS</b>	Gene OMIM Number: 190070	NM ID: NM_004985.3	NO. of Variants:10	Phenotype OMIM Number: 615278
Variant: c.466T>A, c.458A>T, c.455T>G, c.439A>G, c.173C>T, c.101C>T, c.40G>A, c.64C>G, c.178G>C, c.101C>G					
<b>Disease: Cardiofaciocutaneous syndrome</b>	<b>Gene: BRAF</b>	Gene OMIM Number: 164757	NM ID: NM_004333.4	NO. of Variants: 33	Phenotype OMIM Number: 115150
Variant: c.741T>G, c.739T>C, c.1495A>G, c.1743T>A, c.1455G>T, c.2126A>G, c.1914T>G, c.1785T>A, c.1695T>G, c.1502A>G, c.1502A>C, c.1497A>C, c.1399T>G, c.823G>A, c.784C>A, c.721A>C, c.1722C>G, c.1801A>C, c.1799T>G, c.1787G>T, c.1785T>G, c.1783T>C, c.1741A>G, c.1600G>C, c.1574T>C, c.1501G>A, c.1455G>C, c.1442C>A, c.785A>C, c.770A>G, c.736G>C, c.730A>C, c.1406G>A					
<b>Disease: Achondroplasia</b>	<b>Gene: FGFR3</b>	Gene OMIM Number: 134934	NM ID: NM_000142.4	NO. of Variants:6	Phenotype OMIM Number: 100800
Variant: c.1138G>A, c.835A>T, c.1031C>G, c.1138G>C, c.1180A>T, c.1130T>G					
<b>Disease: Campomelic dysplasia with autosomal sex reversal</b>	<b>Gene: SOX9</b>	Gene OMIM Number: 608160	NM ID: NM_000346.3	NO. of Variants:19	Phenotype OMIM Number: 114290
Variant: c.323C>T, c.427T>C, c.432-2A>C, c.685+1G>A, c.788_789insG, c.985_986insG, c.1123C>T, c.1320C>G, c.462C>G, c.236A>C, c.506A>C, c.738delG, c.296delG, c.441delC, c.600delC, c.611delT, c.1372delC, c.442G>T, c.472G>A					

<b>Disease: Acampomelic campomelic dysplasia</b>	<b>Gene: SOX9</b>	<b>Gene OMIM Number: 608160</b>	<b>NM ID: NM_000346.3</b>	<b>NO. of Variants:18</b>	<b>Phenotype OMIM Number: 114290</b>
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**Variants:** c.236A>C, c.506A>C, c.738delG, c.296delG, c.441delC, c.600delC, c.611delT, c.1372delC, c.442G>T, c.473C>T, c.517A>G, c.509C>G, c.472G>A, c.316A>G, c.337A>G, c.495C>G, c.527C>T, c.1514\_1515insC

<b>Disease: Campomelic dysplasia</b>	<b>Gene: SOX9</b>	<b>Gene OMIM Number: 608160</b>	<b>NM ID: NM_000346.3</b>	<b>NO. of Variants:33</b>	<b>Phenotype OMIM Number: 11429</b>
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**Variants:** c.462C>G, c.236A>C, c.506A>C, c.738delG, c.296delG, c.441delC, c.600delC, c.611delT, c.1372delC, c.442G>T, c.473C>T, c.517A>G, c.509C>G, c.338T>C, c.358C>T, c.507C>G, c.122C>A, c.227C>A, c.258\_259insG, c.257G>A, c.349C>T, c.432-2A>G, c.451A>T, c.455G>C, c.583C>T, c.694C>T, c.715dupA, c.736dupC, c.886\_887insC, c.1198G>T, c.1242\_1243insT, c.1456\_1457insG, c.1495\_1496insG

<b>Disease: Tuberous sclerosis II</b>	<b>Gene: TSC2</b>	<b>Gene OMIM Number: 191092</b>	<b>NM ID: NM_000548.3</b>	<b>NO. of Variants:476</b>	<b>Phenotype OMIM Number: 613254</b>
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**Variants:** c.139\_140delGA, c.488\_489delTT, c.1565\_1566delAC, c.2071delC, c.3206\_3207delTG, c.4115\_4116delTG, c.4180\_4181delCT, c.4207delG, c.4422\_4423delAG, c.4544delA, c.4642delC, c.4934\_4935delTT, c.5259delG, c.45delT, c.219delT, c.415delA, c.440\_441delCA, c.462delC, c.529delC, c.633delC, c.703\_704delAG, c.729\_730delCT, c.799delC, c.909\_910delCT, c.976delG, c.1002\_1003delGT, c.1143delG, c.1185\_1186delTG, c.1331delA, c.1391delA, c.1410delC, c.1491delC, c.1642\_1643insA, c.1693delC, c.1796delA, c.2182delT, c.2395delC, c.2539delC, c.2639delA, c.2643delT, c.2690delT, c.2784delC, c.3128delA, c.3214delA, c.3294\_3295insC, c.3319\_3320insG, c.3337\_3338insG, c.3351delG, c.3390delC, c.3401delG, c.3520delC, c.3574delC, c.3984delG, c.4279delA, c.4473delA, c.4472\_4473delAA, c.4504delC, c.4578delC, c.4591delG, c.4704delC, c.4710delG, c.4717\_4718insG, c.4762\_4763insC, c.4925delG, c.4938\_4939delGT, c.5024delC, c.5074\_5075insG, c.5136\_5137delCC, c.5170\_5171delCA, c.5173\_5174insG, c.5422\_5423delTG, c.45\_46insT, c.109\_110insG, c.832\_833insC, c.871\_872insC, c.910\_911insCT, c.1628\_1629insC, c.2090\_2091insT, c.2295\_2296insCC, c.2709\_2710insC, c.2717\_2718insA, c.2784\_2785insC, c.4442\_4443insA, c.4991\_4992insG, c.5247\_5248insG, c.5408\_5409insT, c.478C>G, c.1599G>C, c.2197C>G, c.4807G>T, c.4942A>T, c.5160+4A>T, c.224A>G, c.849-3T>G, c.976-15G>A, c.976-3C>G, c.1001T>C, c.1001T>G, c.1347G>C, c.1789C>T, c.1796A>T, c.1831C>T, c.1864C>T, c.2114T>A, c.2150T>G, c.2198T>C, c.2713C>G, c.2726T>A, c.2747T>G, c.3082G>A, c.3106T>C, c.3178T>C, c.3182T>C, c.3596T>G, c.3608C>A, c.3884A>T, c.4489C>A, c.4490C>G, c.4601T>A, c.4604A>C, c.4643T>C, c.4646A>G, c.4700G>A, c.4708A>T, c.4711T>A, c.4735G>A, c.4751T>G, c.4780C>A, c.4787G>T, c.4790T>C, c.4828T>G, c.4859A>G, c.4868C>T, c.4928A>T, c.4929C>G, c.4937T>G, c.4949A>G, c.4952A>G, c.4957T>C, c.5017G>T, c.5018T>A, c.5057A>C, c.5126C>G, c.5160+5G>T, c.5138G>A, c.139-1G>A, c.500G>A, c.826\_827delAT, c.1111C>T, c.1119+1G>C, c.1474C>T, c.1760\_1761delAT, c.1946+1G>A, c.3180G>A, c.3797dupT, c.3999C>A, c.4183C>T, c.5160+1G>T, c.5161-2A>G, c.100A>T, c.115delA, c.139-2A>G, c.169delC, c.187delC, c.266T>A, c.334C>T, c.337-2A>G, c.357delC, c.501G>A, c.509\_510insAT, c.516\_517delGT, c.602delT, c.606delC, c.644\_645delTA, c.648+1G>T, c.826delA, c.855C>A, c.899\_900insT, c.908T>C, c.949\_950insTA, c.1008T>G, c.1094\_1095delTC, c.1228\_1229insG, c.1323G>A, c.1361+1G>A, c.1407delG, c.1528C>T, c.1599+2T>A, c.1616delT, c.1839+1G>A, c.1960\_1961delGG, c.1976\_1977insA, c.2074delG, c.2157T>A, c.2163delG, c.2235\_2236delGA, c.2240\_2241delTG, c.2417\_2418dupTG, c.2433delC, c.2452dupA, c.2530delC, c.2545+2T>C, c.2639+1G>T, c.2649\_2650dupGT, c.2670delT, c.2737\_2738delAC, c.2766delG, c.2785delG, c.2814\_2815delTA, c.2816\_2817dupGT, c.3097\_3098dupTA, c.3099delC, c.3131G>C, c.3131+2T>C, c.3240dupA, c.3312\_3313delGA, c.3316A>T, c.3383delT, c.3397+1G>A, c.3581G>A, c.3599G>C, c.3611-1delG, c.3695delC, c.3755C>A, c.3884-1G>A, c.3982dupA, c.4013\_4014delCA, c.4014dupA, c.4027G>T, c.4030G>T, c.4051G>T, c.4176\_4177delGA, c.4235\_4236delCT, c.4236dupT, c.4255delC, c.4264delA, c.4344dupC, c.4537G>T, c.4569+2T>C, c.4630A>T, c.4662+1G>T, c.4663-2\_4663-1delAG, c.4743delC, c.4854delC, c.4870\_4871delCT, c.4909\_4910delAA, c.5025delG, c.5160+2T>G, c.5161delA, c.5402\_5403delAG, c.3G>A, c.34A>T, c.138+1G>A, c.138+2T>C, c.225+2T>A, c.226-2A>G, c.246G>A, c.268C>T, c.336+1G>A, c.336+2T>G, c.337-1G>T, c.403dupG, c.451A>T, c.465C>G, c.469G>T, c.496C>T, c.569dupA, c.598C>T, c.600-2A>G, c.600-1G>A, c.618C>A, c.648+1G>A, c.684C>A, c.707dupT, c.731G>A, c.759C>A, c.760G>T, c.767dupG, c.768C>A, c.770G>A, c.774+2T>A, c.775-1G>A, c.848+1G>A, c.849-2A>T, c.849-1G>A, c.880G>A, c.911G>A, c.972C>G, c.975+1G>T, c.976-1G>A, c.979\_980dupAT, c.1042A>T, c.1060C>T, c.1096G>T, c.1108C>T, c.1117C>T, c.1249C>T, c.1255C>T, c.1257+2T>G, c.1258-2A>C, c.1258-1G>A, c.1322G>A, c.1327C>T, c.1336C>T, c.1348G>T, c.1362-2A>G, c.1362-1G>A, c.1372C>T, c.1432C>T, c.1444-2A>C, c.1444-1G>A, c.1447G>T, c.1472C>A, c.1507C>T, c.1513C>T, c.1563dupA, c.1599+1G>A, c.1599+2T>C, c.1661C>A, c.1714C>T, c.1783C>T, c.1790A>G, c.1792T>C, c.1794C>G, c.1820C>A, c.1832G>A, c.1839+1G>T, c.1840-2A>T, c.1841C>A, c.1844T>C, c.1901\_1902insAT, c.1942dupT, c.1947-2A>G, c.1965dupT, c.1969A>T, c.2083C>T, c.2087\_2088insAA, c.2098-2A>G, c.2098G>A, c.2103dupT, c.2106dupC, c.2108G>A, c.2109G>A, c.2113G>A, c.2158A>T, c.2194C>T, c.2221-2A>C, c.2225C>G, c.2251C>T, c.2328C>A, c.2353C>T, c.2355+1G>T, c.2355+2T>C, c.2356-2A>C, c.2356-1G>A, c.2370C>G, c.2375T>G, c.2404dupA, c.2410T>C, c.2430dupC, c.2461A>T, c.2489T>G, c.2491\_2492insTA, c.2531T>G, c.2540T>C, c.2546-2A>G, c.2546-2A>T, c.2590C>T, c.2639+1G>C, c.2661T>A, c.2665G>C, c.2666C>T, c.2687G>A, c.2688G>A, c.2690T>C, c.2713C>T, c.2714G>A, c.2742+1G>A, c.2743-2A>C, c.2743-2A>G, c.2743-1G>C, c.2768dupC, c.2785G>T, c.2824G>T, c.2837+1G>T, c.2974C>T, c.3028C>T, c.3076dupT, c.3094C>T, c.3095G>C, c.3098dupA, c.3099C>G, c.3099C>A, c.3131+1G>A, c.3132-2A>C, c.3203C>T, c.3212C>G, c.3265C>T, c.3284+1G>C, c.3355C>T, c.3397+2T>G, c.3412C>T, c.3425dupT, c.3442C>T, c.3532C>T, c.3574C>T, c.3575dupA, c.3598C>T, c.3610+1G>A, c.3610+1G>C, c.3662C>A, c.3685C>T, c.3696dupT, c.3750C>G, c.3755C>G, c.3884-1G>C, c.4005+1G>T, c.4037C>A, c.4096G>T, c.4129C>T, c.4255C>T, c.4289G>A, c.4298C>A, c.4318C>T, c.4375C>T, c.4397C>A, c.4438\_4439insA, c.4439T>A, c.4493G>C, c.4493G>A, c.4494-1\_4494insC, c.4507C>T, c.4515C>A, c.4573C>T, c.4606C>T, c.4620C>G, c.4620C>A, c.4662G>T, c.4663-1G>A, c.4672G>A, c.4700G>T, c.4713C>G, c.4744dupA, c.4779C>G, c.4813C>T, c.4830G>A, c.4846C>T, c.4850-2A>C, c.4850-1G>A, c.4858C>T, c.4871T>C, c.4918C>T, c.4925G>A, c.4989+1G>A, c.4989+1\_4989+2insA, c.4993C>T, c.5024C>T, c.5032dupT, c.5034C>G, c.5043C>G, c.5069-2A>G, c.5069-1G>A, c.5069-1G>T, c.5112dupT, c.5160+1G>A, c.5160+1\_5160+2insT, c.5161-1G>A, c.5161-1G>C, c.5161-1G>T, c.5170C>T, c.5208C>A, c.5220G>A, c.5227C>T, c.5228G>A, c.5388dupC

<b>Disease: Tuberous sclerosis I</b>	<b>Gene: TSC1</b>	<b>Gene OMIM Number: 605284</b>	<b>NM ID: NM_000368.4</b>	<b>NO. of Variants:176</b>	<b>Phenotype OMIM Number: 191100</b>
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**Variants:** c.988\_989delCT, c.90delA, c.901\_902delCA, c.834\_835delTC, c.831\_832delGT, c.749delIT, c.749\_750insT, c.723\_724insA, c.658delIG, c.654\_655insA, c.648delIT, c.647\_648delTT, c.63\_64insTG, c.563\_564delTT, c.433\_434delICA, c.2701\_2702delAG, c.2672delA, c.2672\_2673insA, c.2569delIG, c.2569\_2570insG, c.252\_253insC, c.2510\_2511insA, c.2501\_2502insA, c.2470\_2471delAC, c.2364delA, c.2342\_2343insAA, c.2319delIC, c.2175\_2176insA, c.2145delIG, c.2109\_2110delCT, c.195delA, c.1907\_1908delAG, c.1904\_1905delCA, c.1841delA, c.1823\_1824delIT, c.1792\_1793insAA, c.1788delIT, c.1697delC, c.1656\_1657insT, c.1551delIG, c.1516\_1517insTC, c.1490\_1491delITG, c.148delC, c.1434delA, c.1425delA, c.1271\_1272delGA, c.1257delC, c.1020delA, c.1015\_1016insA, c.2177C>A, c.1030-3C>G, c.671T>G, c.611G>C, c.473T>C, c.350T>C, c.2530C>T, c.2272C>T, c.2111\_2112delAT, c.1781delIT, c.1708\_1709delAG, c.1580\_1581delAG, c.1530\_1531delCA, c.1453G>T, c.2698\_2699delCA, c.2668A>T, c.2582delIT, c.2503-2delA, c.2287C>T, c.2263C>T, c.2155delC, c.2022delC, c.1997+1G>C, c.1966G>T, c.1964delA, c.1958dupT, c.1955\_1956delITG, c.1846delIG, c.1799delA, c.1615delIT, c.1560delC, c.1533delIT, c.1250delC, c.1203delIT, c.1119C>G, c.1027C>T, c.989delIT, c.988delC, c.913+1G>A, c.893dupC, c.801dupA, c.745delA, c.591dupC, c.585C>A, c.562T>G, c.533dupT, c.527\_528delAT, c.508+1G>A, c.433C>T, c.338T>A, c.203\_204delAT, c.70delG, c.2698C>T, c.2692C>T, c.2689C>T, c.2650dupT, c.2503-1G>C, c.2503-1G>T, c.2503-2A>G, c.2497C>T, c.2389C>T, c.2362G>T, c.2356C>T, c.2332C>T, c.2299C>T, c.2293C>T, c.2283C>G, c.2283C>A, c.2250G>A, c.2227C>T, c.2074C>T, c.2042-2A>G, c.2041+2T>C, c.2028G>A, c.1963C>T, c.1883T>A, c.1773\_1774dupGA, c.1759A>T, c.1729G>T, c.1717C>T, c.1676dupG, c.1525C>T, c.1498C>T, c.1439-2A>G, c.1432\_1433dupGA, c.1411\_1412insT, c.1331C>G, c.1118dupA, c.1030-1G>A, c.1001C>A, c.989dupT, c.988dupC, c.982C>T, c.936C>A, c.901C>T, c.891T>G, c.814G>T, c.772G>T, c.749T>G, c.745A>T, c.737+1G>A, c.733C>T, c.709dupT, c.682C>T, c.664-1G>A, c.647T>C, c.572T>G, c.572T>A, c.569G>C, c.539T>C, c.495C>A, c.397G>T, c.395G>A, c.372dupT, c.325C>T, c.309G>A, c.308G>A, c.278T>G, c.272C>A, c.215T>C, c.211-1G>A, c.210+1G>A, c.196G>T, c.182T>G, c.182T>C, c.149T>C, c.107-2A>G

<b>Disease: Osteogenesis imperfecta, type I</b>	<b>Gene: COL1A1</b>	<b>Gene OMIM Number: 120150</b>	<b>NM ID: NM_000088.3</b>	<b>NO. of Variants:322</b>	<b>Phenotype OMIM Number: 166200</b>
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**Variants:** c.809delIT, c.809\_810insT, c.768\_769insC, c.700delIG, c.671delIG, c.614delC, c.569delC, c.540\_541insC, c.533delC, c.484delC, c.4332\_4333insC, c.3765delC, c.3749delIG, c.3699\_3700insC, c.3638delIG, c.3584delIG, c.3560delIG, c.3540delC, c.3402delC, c.3371delIG, c.3258delC, c.3168\_3169insC, c.3065delIG, c.3026delC, c.3008delC, c.3008\_3009insC, c.2774delC, c.2732delIG, c.2684delC, c.2612delC, c.2522delC, c.246delC, c.2450\_2451insC, c.2444delIG, c.2426\_2427insG, c.2424delC, c.2390delIG, c.2366delC, c.2321delC, c.2097delC, c.2084delIG, c.2072delC, c.2037\_2038delAG, c.2016\_2017insC, c.1994delIG, c.1920\_1921insC, c.1893\_1894delAG, c.1886delIG, c.1865delC, c.1814delIG, c.1787delIG, c.1667delC, c.1667\_1668insC, c.1661delIG, c.157\_158delITG, c.1486\_1487delCC, c.1379delC, c.1292delIG, c.1269delC, c.1265delIG, c.1261delC, c.1251delC, c.1208delC, c.1127delC, c.1821+1G>A, c.1066G>T, c.994G>A, c.2299G>A, c.3262G>T, c.2523delIT, c.4009G>T, c.3208-1G>C, c.3045+1G>A, c.2089C>T, c.1057-1G>T, c.1057-2A>G, c.1003-1G>C, c.1003-2A>G, c.642+5G>A, c.299-2A>G, c.904-9G>A, c.3470G>A, c.3263G>A, c.3208G>A, c.788G>T, c.588+5G>A, c.543+5G>A, c.3825G>A, c.3360delIT, c.3207+1\_3207+2delITG, c.3162delIT, c.2991delIT, c.2784delIT, c.2775delIT, c.2010delIT, c.1812delIT, c.1452delIT, c.1128delIT, c.1057-1G>A, c.1002+2T>C, c.751-1G>A, c.579delIT, c.148C>T, c.1A>G, c.4249-2delA, c.4156A>T, c.4054delC, c.4051C>T, c.3766delIG, c.3718C>T, c.3589\_3590delGA, c.3531+1G>C, c.3369+1G>C, c.3328delC, c.3313delA, c.3309delC, c.3208-1G>A, c.3201delIT, c.3079delIG, c.2646\_2647delAG, c.2614-1G>A, c.2451delIT, c.2329delIG, c.2236-1G>A, c.2235\_2235+1delAG, c.2085\_2086delITC, c.1983+2T>G, c.1944delIT, c.1900C>T, c.1876-2delA, c.1858G>T, c.1668delIT, c.1608delIT, c.1201\_1202delGG, c.1200+1\_1200+2delITG, c.1200+1G>C, c.1155+2T>G, c.1128\_1129insC, c.1065delIT, c.982\_983dupAC, c.775delIG, c.697-2delA, c.669delA, c.358C>T, c.143dupA, c.67C>T, c.38T>G, c.4338dupC, c.4021C>T, c.4005+1G>C, c.4005+1G>A, c.3975G>A, c.3974G>A, c.3925C>T, c.3838C>T, c.3815-2A>G, c.3814+2T>C, c.3814+2T>G, c.3814+1G>C, c.3807G>A, c.3607C>T, c.3532-1G>A, c.3531+2T>C, c.3531+1G>A, c.3423+1G>A, c.3421C>T, c.3385C>T, c.3369+1G>A, c.3369+1G>T, c.3285dupC, c.3262-1G>A, c.3207+2T>C, c.3207+1G>A, c.3207+1G>C, c.3100-1G>A, c.3099+1G>A, c.3076C>T, c.3046-2A>G, c.3045+1G>T, c.2938-1G>A, c.2938-2A>G, c.2869C>T, c.2668-1G>A, c.2667+1G>A, c.2594dupG, c.2560-1G>A, c.2559+2T>A, c.2550dupT, c.2464C>T, c.2452-1G>A, c.2452-2A>G, c.2452-2A>T, c.2451+1G>A, c.2451+1G>T, c.2398-1G>A, c.2397\_2397+1insC, c.2344-1G>A, c.2343+1G>A, c.2236-2A>G, c.2128-1G>A, c.2127+2T>G, c.2127+1G>A, c.2032G>T, c.2028+2T>G, c.1984-1G>A, c.1983+1G>A, c.1930-1G>A, c.1930-1G>C, c.1930-2A>G, c.1929+1G>C, c.1929+1G>A, c.1876-1G>A, c.1875+1G>A, c.1821+1G>C, c.1821+1G>T, c.1768-1G>A, c.1718\_1719insAC, c.1615-1G>C, c.1615-1G>T, c.1614+1G>C, c.1614+1G>A, c.1516-1G>C, c.1516-1G>T, c.1516-2A>G, c.1461+1G>A, c.1414C>T, c.1354-1G>A, c.1354-1G>T, c.1354-2A>G, c.1353+2T>C, c.1353+1G>C, c.1300-1G>A, c.1299+1G>A, c.1201-1G>A, c.1200+1G>T, c.1156-1G>A, c.1155+1G>A, c.1099C>T, c.1081C>T, c.1057-2A>T, c.983\_984dupCT, c.958-1G>A, c.858+1G>A, c.858+1G>T, c.757C>T, c.751-2A>G, c.697-1G>A, c.697-2A>G, c.697-2A>T, c.696+2T>G, c.696+1G>A, c.589-1G>A, c.589-2A>G, c.588+1G>A, c.543+2T>A, c.543+2T>C, c.543+1G>T, c.472-2A>G, c.370-1G>A, c.334-2A>G, c.333+2T>C, c.333+1G>A, c.333+1G>T, c.299-1G>A, c.299-1G>C, c.141C>A, c.104-1G>T, c.103+1G>A, c.103+1G>C, c.2T>G, c.2T>C, c.3790A>G, c.2644C>T, c.2559+1G>A, c.2155G>A, c.2028+1G>A, c.1984-2A>C, c.1155+1G>C, c.1056+1G>A, c.859-2A>C, c.4166T>C, c.3380G>A, c.3290G>T, c.4321G>T, c.3505G>A, c.769G>A, c.3567delIT, c.3495delIT, c.2685delIT, c.517G>T, c.3987delC, c.3531+1G>T, c.3477delIT, c.3450dupT, c.3241delIG, c.3135delIT, c.3114delIG, c.3027delIT, c.2410G>T, c.1930-2A>C, c.1866delIT, c.898C>T, c.804+2\_804+3delITG, c.750+2T>A, c.484C>T, c.268G>T, c.253G>T, c.189C>A, c.144delIT, c.4006-1G>A, c.3910C>T, c.3349C>T, c.2533G>A, c.2451\_2451+1insT, c.1696G>A, c.1405C>T, c.1200+1G>A, c.967G>T, c.750+1G>A, c.697-1G>C, c.104-1G>A, c.671G>A

<b>Disease: Osteogenesis imperfecta, type II</b>	<b>Gene: COL1A1</b>	<b>Gene OMIM Number: 120150</b>	<b>NM ID: NM_000088.3</b>	<b>NO. of Variants:32</b>	<b>Phenotype OMIM Number: 166210</b>
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**Variants:** c.1102G>A, c.760G>A, c.4163T>G, c.3515G>A, c.3496G>T, c.3073G>A, c.2605G>T, c.2317G>T, c.1966G>A, c.957+5G>A, c.64G>C, c.4247delC, c.4309delC, c.4105delC, c.4265G>A, c.3969dupT, c.3424-1G>C, c.3423+2T>A, c.3370G>T, c.3261+2T>C, c.3261+1G>A, c.3100-2A>C, c.3100-2A>G, c.2228G>T, c.2227G>A, c.1767+2T>C, c.1705G>C, c.1669-2A>G, c.1668+1G>A, c.957+2T>A, c.957+1G>T, c.2235+1G>A

<b>Disease: Osteogenesis imperfecta, type III</b>	<b>Gene: COL1A1</b>	<b>Gene OMIM Number: 120150</b>	<b>NM ID: NM_000088.3</b>	<b>NO. of Variants:45</b>	<b>Phenotype OMIM Number: 259420</b>
<b>Variants:</b> c.671G>A, c.1243C>T, c.2515G>A, c.1777G>T, c.608G>T, c.1121G>C, c.4375G>C, c.4391T>C, c.2937+4A>C, c.1976G>C, c.696+3A>C, c.3540_3541insC, c.3580_3581delGC, c.3277delC, c.2786delG, c.2268_2269delTC, c.2208delT, c.1822-2delA, c.1822-2A>T, c.1631delC, c.1072delC, c.904-1G>A, c.630delG, c.4308_4309insA, c.4248+1G>A, c.3208-2A>C, c.2075G>C, c.2065G>A, c.1875+1G>C, c.1669-1G>C, c.750+1G>C, c.1102G>A, c.760G>A, c.1821+1G>A, c.994G>A, c.2299G>A, c.2644C>T, c.2559+1G>A, c.2155G>A, c.2028+1G>A, c.1984-2A>C, c.1155+1G>C, c.1056+1G>A, c.859-2A>C, c.2235+1G>A					
<b>Disease: Osteogenesis imperfecta, type IV</b>	<b>Gene: COL1A1</b>	<b>Gene OMIM Number: 120150</b>	<b>NM ID: NM_000088.3</b>	<b>NO. of Variants:48</b>	<b>Phenotype OMIM Number: 166220</b>
<b>Variants:</b> c.4364G>T, c.3853G>C, c.281T>A, c.1579G>T, c.588+4A>T, c.4352dupA, c.4291_4292insA, c.2250delC, c.1897G>T, c.1822-2A>G, c.695delA, c.588+1G>T, c.103+2T>C, c.3897C>A, c.3824G>A, c.3806G>A, c.2128-1G>C, c.1876-1G>C, c.1876-2A>G, c.1461+1G>A, c.1379_1380insC, c.1299+1G>C, c.859-1G>A, c.750+1G>T, c.697-1G>T, c.642+1G>A, c.642+1G>T, c.1243C>T, c.2515G>A, c.1777G>T, c.608G>T, c.1066G>T, c.994G>A, c.3008delC, c.2684delC, c.3262G>T, c.2523delT, c.1667delC, c.4009G>T, c.3208-1G>C, c.3045+1G>A, c.2089C>T, c.1057-1G>T, c.1057-2A>G, c.1003-1G>C, c.1003-2A>G, c.642+5G>A, c.299-2A>G					
<b>Disease: Osteogenesis imperfecta, type I</b>	<b>Gene: COL1A2</b>	<b>Gene OMIM Number: 120160</b>	<b>NM ID: NM_000089.3</b>	<b>NO. of Variants:18</b>	<b>Phenotype OMIM Number: 166200</b>
<b>Variants:</b> c.433-1G>C, c.486+1G>C, c.693+1G>A, c.3304G>T, c.4040T>A, c.432+5G>A, c.3089G>C, c.1298G>A, c.12T>G, c.3773G>A, c.595-2A>G, c.1361G>T, c.2188G>T, c.2565+1G>A, c.2835+1G>A, c.3105+2T>C, c.4060C>T, c.577G>A					
<b>Disease: Osteogenesis imperfecta, type II</b>	<b>Gene: COL1A2</b>	<b>Gene OMIM Number: 120160</b>	<b>NM ID: NM_000089.3</b>	<b>NO. of Variants:9</b>	<b>Phenotype OMIM Number: 166210</b>
<b>Variants:</b> c.1863G>A, c.1316G>A, c.1532G>A, c.1720-2A>G, c.2673+1G>A, c.2944-2A>G, c.1433G>A, c.2386G>A, c.3008G>A					
<b>Disease: Osteogenesis imperfecta, type III</b>	<b>Gene: COL1A2</b>	<b>Gene OMIM Number: 120160</b>	<b>NM ID: NM_000089.3</b>	<b>NO. of Variants:12</b>	<b>Phenotype OMIM Number: 259420</b>
<b>Variants:</b> c.982G>A, c.1406G>C, c.3350A>G, c.1035_1035+1delTG, c.792+2T>A, c.1099G>T, c.1162G>C, c.1937G>T, c.1433G>A, c.2386G>A, c.3008G>A, c.3089G>C					
<b>Disease: Osteogenesis imperfecta, type IV</b>	<b>Gene: COL1A2</b>	<b>Gene OMIM Number: 120160</b>	<b>NM ID: NM_000089.3</b>	<b>NO. of Variants:19</b>	<b>Phenotype OMIM Number: 166220</b>
<b>Variants:</b> c.3527G>T, c.694-2A>C, c.486+1G>T, c.694-2A>G, c.594+2T>G, c.792+1G>A, c.792+1G>T, c.792+2T>C, c.793G>C, c.1009G>A, c.2027G>T, c.2297G>T, c.3487T>C, c.3944A>T, c.3952dupT, c.982G>A, c.1406G>C, c.3350A>G, c.433-1G>C					
<b>Disease: Hyper-IgE recurrent infection syndrome</b>	<b>Gene: STAT3</b>	<b>Gene OMIM Number: 102582</b>	<b>NM ID: NM_139276.2</b>	<b>NO. of Variants:45</b>	<b>Phenotype OMIM Number: 147060</b>
<b>Variants:</b> c.2147C>T, c.1699A>G, c.1414A>G, c.1110-3C>A, c.2134T>C, c.1593A>T, c.1281+1delG, c.1276T>C, c.995A>C, c.2137G>C, c.2132T>C, c.2129T>G, c.2125A>G, c.2116C>A, c.1994T>A, c.1979T>C, c.1974G>C, c.1970A>C, c.1913A>G, c.1909G>A, c.1865C>T, c.1863C>G, c.1858A>G, c.1850G>A, c.1591A>G, c.1407G>T, c.1406A>G, c.1310A>C, c.1309C>T, c.1294G>C, c.1268G>A, c.1166C>T, c.1152T>A, c.1151T>G, c.1151T>C, c.1145G>A, c.1144C>G, c.1144C>T, c.1140-2A>G, c.1139+1G>T, c.1110-1G>T, c.1110-2A>G, c.1025G>A, c.995A>T, c.994C>T					
<b>Disease: Stickler syndrome, type I</b>	<b>Gene: COL2A1</b>	<b>Gene OMIM Number: 120140</b>	<b>NM ID: NM_001844.4</b>	<b>NO. of Variants:157</b>	<b>Phenotype OMIM Number: 108300</b>
<b>Variants:</b> c.996_997delAG, c.941delC, c.883delC, c.724delC, c.583_584insC, c.509_510insC, c.406_407insG, c.3906delC, c.3864_3865delCT, c.3641_3642insC, c.3623delC, c.3464_3465insG, c.3401delG, c.3356delG, c.3325delC, c.3137_3138insC, c.311delG, c.3012delC, c.2999delC, c.2976_2977delAG, c.294delG, c.2858delC, c.2813delC, c.2789delG, c.2787delA, c.2723_2724insC, c.2715_2716insT, c.2678delC, c.2678_2679insC, c.2539delG, c.2478_2479delGA, c.2263_2264delAG, c.1970delG, c.1943_1944insG, c.1931delC, c.1821delC, c.1783_1784insG, c.1732delT, c.166_167delGT, c.1602_1603insG, c.146delC, c.1416delA, c.1313delG, c.1172delC, c.3508G>A, c.3624delT, c.3138delT, c.2818C>T, c.2814delT, c.2382delT, c.4074+2_4074+3delTG, c.3872_3873delCT, c.3778_3779delAA, c.3495delT, c.3488delG, c.3419G>C, c.3228delT, c.2751delT, c.2471delG, c.2302-1G>A, c.2077_2078delGG, c.1995+1G>A, c.1963G>T, c.1908delT, c.1828delG, c.1674delA, c.1522G>T, c.1221+1delG, c.1095delT, c.1032delA, c.933delT, c.793delG, c.744delT, c.572delC, c.510delT, c.237delG, c.85C>T, c.4317+2T>C, c.4074+1G>T, c.3886+2T>C, c.3886+1G>A, c.3886+1G>T, c.3878G>A, c.3714C>A, c.3709C>T, c.3597+2T>G, c.3597+1G>A, c.3596dupC, c.3574C>T, c.3419G>A, c.3327+1G>C, c.3280C>T, c.3274-2A>G, c.3165+1G>T, c.3112-1G>A, c.3111+1G>T, c.3003+1G>A, c.3003+1G>T, c.2896-1G>A, c.2869G>T, c.2839C>T, c.2794C>T, c.2653G>T, c.2626-2A>G, c.2625+1G>T, c.2518-1G>A, c.2517+2T>G, c.2517+1G>C, c.2493dupA, c.2473G>T, c.2467G>T, c.2436dupA, c.2355+1G>A, c.2353C>T, c.2194-1G>A, c.2193+2T>C, c.2101C>T, c.2095-1G>A, c.2094+1G>A, c.2050-1G>C, c.2049+1G>A, c.1996-1G>A, c.1957C>T, c.1942-2A>G, c.1941+1G>A, c.1888-1G>C, c.1888-2A>G, c.1887+1G>A, c.1833+1G>A, c.1777C>T, c.1735-1G>A, c.1714C>T, c.1693C>T, c.1528-2A>G, c.1527+1G>C, c.1399_1400dupCA, c.1366-2A>T, c.1222-2A>G, c.1221+1G>A, c.1221+1G>C, c.1123-1G>A, c.1069-2A>G, c.1030C>T, c.870+1G>A, c.756dupT, c.655-1G>C, c.654+1G>A, c.625C>T, c.556G>T, c.430-1G>C, c.342+1G>A, c.293-1G>A, c.258C>A, c.192C>A, c.123T>A, c.3G>T, c.1962C>T					

<b>Disease: Stickler syndrome, type II</b>	<b>Gene: COL11A1</b>	<b>Gene OMIM Number: 120280</b>	<b>NM ID: NM_001854.3</b>	<b>NO. of Variants:17</b>	<b>Phenotype OMIM Number: 604841</b>
<b>Variant:</b> c.3168+5G>A, c.3816+2_3816+3insT, c.3816+1G>A, c.3168+1G>T, c.1630-2delA, c.2808+1G>C, c.2043+1G>T, c.1845+1G>A, c.1191delT, c.4554+1G>A, c.4519-2A>G, c.3978+1G>A, c.3762+1G>A, c.2043+1G>A, c.1900-1G>A, c.1681C>T, c.1245+1G>A					
<b>Disease: Pfeiffer syndrome</b>	<b>Gene: FGFR2</b>	<b>Gene OMIM Number: 176943</b>	<b>NM ID: NM_000141.4</b>	<b>NO. of Variants:8</b>	<b>Phenotype OMIM Number: 101600</b>
<b>Variant:</b> c.1988G>A, c.1198A>G, c.1922A>G, c.1646A>C, c.1024T>G, c.940-1G>A, c.940-2A>T, c.870G>C					
<b>Disease: Pfeiffer syndrome</b>	<b>Gene: FGFR1</b>	<b>Gene OMIM Number: 136350</b>	<b>NM ID: FGFR1</b>	<b>NO. of Variants:1</b>	<b>Phenotype OMIM Number: 101600</b>
<b>Variant:</b> c.755C>G					
<b>Disease: Jackson-Weiss syndrome</b>	<b>Gene: FGFR2</b>	<b>Gene OMIM Number: 176943</b>	<b>NM ID: NM_000141.4</b>	<b>NO. of Variants:2</b>	<b>Phenotype OMIM Number: 123150</b>
<b>Variant:</b> c.958_959delAC, c.1040C>G					
<b>Disease: Hutchinson-Gilford progeria</b>	<b>Gene: LMNA</b>	<b>Gene OMIM Number: 150330</b>	<b>NM ID: NM_170707.3</b>	<b>NO. of Variants:1</b>	<b>Phenotype OMIM Number: 176670</b>
<b>Variant:</b> c.1824C>T					
<b>Disease: Crouzon syndrome with acanthosis nigricans</b>	<b>Gene: FGFR3</b>	<b>Gene OMIM Number: 134934</b>	<b>NM ID: NM_000142.4</b>	<b>NO. of Variants:1</b>	<b>Phenotype OMIM Number: 612247</b>
<b>Variant:</b> c.1172C>A					
<b>Disease: Crouzon syndrome</b>	<b>Gene: FGFR2</b>	<b>Gene OMIM Number: 176943</b>	<b>NM ID: NM_000141.4</b>	<b>NO. of Variants:19</b>	<b>Phenotype OMIM Number: 123500</b>
<b>Variant:</b> c.1024T>C, c.1021A>C, c.812G>T, c.1040C>G, c.2032A>G, c.184T>C, c.1851G>C, c.1084+1G>T, c.817_818insGAG, c.1645A>C, c.1084+2T>C, c.1084+1G>A, c.1061C>G, c.1025G>A, c.1024T>A, c.923A>G, c.866A>C, c.863T>A, c.799T>C					
<b>Disease: Costello syndrome</b>	<b>Gene: HRAS</b>	<b>Gene OMIM Number: 190020</b>	<b>NM ID: NM_005343.2</b>	<b>NO. of Variants:4</b>	<b>Phenotype OMIM Number: 218040</b>
<b>Variant:</b> c.34G>A, c.350A>G, c.436G>A, c.34G>T					
<b>Disease: CHARGE syndrome</b>	<b>Gene: CHD7</b>	<b>Gene OMIM Number: 608892</b>	<b>NM ID: NM_017780.3</b>	<b>NO. of Variants:560</b>	<b>Phenotype OMIM Number: 214800</b>
<b>Variant:</b> c.718delC, c.1925delA, c.2990delT, c.3572_3573delAA, c.3937delT, c.4634delT, c.7524delC, c.160delC, c.257delC, c.285delG, c.284_285delGG, c.595delT, c.627delG, c.674delC, c.729delC, c.780delC, c.791_792delTC, c.865delA, c.1044delC, c.1168_1169insTT, c.1319delC, c.1374_1375delTC, c.1388delG, c.1544delC, c.1678delG, c.1683delC, c.1686delT, c.1793delA, c.2034delA, c.2118delA, c.2180delT, c.2566delA, c.2585delA, c.2620delG, c.2669_2670delTT, c.2735delC, c.2859delG, c.2908delG, c.3165delC, c.3318delA, c.3322delC, c.3339delG, c.3359delG, c.3377_3378insT, c.3548delA, c.3693delA, c.3807delT, c.4087delC, c.4171delC, c.4183delC, c.4226_4227delTG, c.4253delA, c.4257delC, c.4270delA, c.4402delG, c.4517_4518insG, c.4527delT, c.4656_4657insT, c.4686delC, c.5054delT, c.5097_5098insA, c.5250delA, c.5499_5500delAA, c.5574delA, c.5588delC, c.5680_5681delAG, c.5776delA, c.5800delC, c.5932delG, c.5991delT, c.6018delA, c.6044delT, c.6224delG, c.6304delG, c.6320_6321delAC, c.6393delT, c.6405_6406delAG, c.6461delC, c.6667delG, c.6705delA, c.6998delC, c.7027delC, c.7180delC, c.7184_7185delCT, c.7219delA, c.7231delG, c.7320delA, c.7384delT, c.7400delT, c.7577delT, c.7650_7651delGA, c.7782delG, c.7895delA, c.7921_7922delTT, c.8279delA, c.8459_8460insCT, c.8491delT, c.8507delC, c.8565delA, c.327_328insC, c.900_901insC, c.1247_1248insG, c.1730_1731insA, c.1865_1866insG, c.1925_1926insA, c.1936_1937insAA, c.1953_1954insA, c.1989_1990insAA, c.1989_1990insA, c.2438_2439insA, c.2571_2572insA, c.2577_2578insT, c.2689_2690insC, c.3122_3123insT, c.3573_3574insA, c.3728_3729insA, c.4112_4113insT, c.4667_4668insC, c.4723_4724insG, c.4850_4851insG, c.5069_5070insC, c.5332_5333insC, c.5564_5565insC, c.5636_5637insA, c.5910_5911insA, c.6018_6019insA, c.6169_6170insC, c.6857_6858insG, c.7227_7228insT, c.7593_7594insG, c.7655_7656insC, c.2096G>A, c.2219A>G, c.2613G>T, c.2957+5G>A, c.3202-3T>G, c.3378+5G>C, c.3952T>C, c.4353+3A>G, c.4406A>G, c.4644+5G>A, c.5050+3A>T, c.5390G>T, c.5405-17G>A, c.5405-7G>A, c.5534G>A, c.6103+5G>C, c.6271T>C, c.6857G>C, c.7165-4A>G, c.8077-10T>A, c.232C>T, c.934C>T, c.1714C>T, c.6272G>A, c.282delT, c.3209delT, c.3490C>T, c.3526C>T, c.5210+2T>C, c.5405-2A>G, c.6243C>A, c.6892C>T, c.77_78delAA, c.219dupT, c.222delG, c.235A>T, c.317delA, c.360delC, c.378C>A, c.635delA, c.669_670insG, c.781delT, c.785delC, c.799G>T, c.808delG, c.921_922delAG, c.959_960delAG, c.964_965delTT, c.1141_1142delAT, c.1152delA, c.1295delA, c.1528delC, c.1665+1G>C, c.1683_1684delCT, c.1740delA, c.1918delG, c.2049delG, c.2145delC, c.2157delA, c.2236delC, c.2238delG, c.2238+2T>A, c.2244_2245delAC, c.2362C>T, c.2443-1delG, c.2464G>T, c.2490C>A, c.2499-2A>C, c.2517delG, c.2706_2707delTC, c.2739delT, c.2829delG, c.2836-2A>G, c.2886T>G, c.2905_2906delAG, c.2916_2917delGT, c.2966delG, c.3023_3024delAT, c.3138delT, c.3177T>G, c.3336delC, c.3514_3515delGA, c.3640C>T, c.3734delT, c.3750delG, c.4012_4013delGG, c.4203_4204delTA, c.4295_4296delTG, c.4354-1G>A, c.4361_4362delAG, c.4393delC, c.4424delA, c.4645-1G>C, c.4665delT, c.4731delA, c.4862G>A, c.5051-1G>A,					

c.5074G>T, c.5592delC, c.5627C>G, c.5656delC, c.5687delC, c.5768dupG, c.5768\_5769delGC, c.6070delC, c.6179delT, c.6209delA, c.6217C>T, c.6228delG, c.6268dupT, c.6326delT, c.6502delC, c.6571delG, c.6712G>T, c.6716delA, c.6746delA, c.6835delG, c.6904delG, c.7047C>A, c.7106delT, c.7234G>T, c.7249delA, c.7328delA, c.7344\_7345delAA, c.7769delA, c.7803delC, c.7875\_7876delGA, c.7884\_7885delTA, c.8267delC, c.151C>T, c.253C>T, c.388C>T, c.406C>T, c.490C>T, c.502C>T, c.511C>T, c.538C>T, c.550C>T, c.562C>T, c.601C>T, c.604C>T, c.608dupA, c.619C>T, c.664C>T, c.689C>G, c.718C>T, c.763C>T, c.804\_805insAT, c.844C>T, c.889C>T, c.925C>T, c.939T>G, c.995T>G, c.1024C>T, c.1036A>T, c.1078G>T, c.1093C>T, c.1095\_1096insTC, c.1116dupA, c.1123C>T, c.1135C>T, c.1153C>T, c.1159C>T, c.1170T>G, c.1190C>G, c.1234C>T, c.1310dupA, c.1312C>T, c.1366C>T, c.1465C>T, c.1480C>T, c.1488dupA, c.1510C>T, c.1576C>T, c.1610G>A, c.1610\_1611insA, c.1615C>T, c.1645C>T, c.1689dupA, c.1735C>T, c.1774C>T, c.1786C>T, c.1818\_1819insAT, c.1940C>G, c.1972G>T, c.1973\_1974insT, c.1983dupG, c.1990G>T, c.2096+2T>A, c.2096+2T>C, c.2097-1G>A, c.2114C>A, c.2181dupA, c.2196dupA, c.2238+1G>A, c.2238+1G>T, c.2238+2T>G, c.2254A>T, c.2311G>T, c.2374C>T, c.2440C>T, c.2443-2A>T, c.2443-2A>G, c.2443-1G>A, c.2498+1G>T, c.2498+1\_2498+2insT, c.2498+2T>C, c.2501C>T, c.2505T>A, c.2505T>G, c.2520G>C, c.2520G>A, c.2572C>T, c.2584A>T, c.2724G>A, c.2737\_2738insTC, c.2753G>A, c.2757dupG, c.2764C>T, c.2815G>T, c.2839C>T, c.2858G>A, c.2957+2T>C, c.2957+2T>G, c.2958-2A>T, c.2958-1G>A, c.2959C>T, c.3024T>G, c.3053\_3054insA, c.3082A>G, c.3089A>G, c.3091T>G, c.3091T>C, c.3106C>T, c.3117dupA, c.3169C>T, c.3173T>A, c.3202-2A>G, c.3202-1G>A, c.3205C>T, c.3245C>A, c.3297G>A, c.3301T>C, c.3379-1G>A, c.3522+2T>C, c.3522+2T>G, c.3523-1G>C, c.3535C>T, c.3616dupA, c.3641A>G, c.3646A>T, c.3654C>G, c.3655C>T, c.3768C>G, c.3770T>G, c.3778+1G>A, c.3779-2A>G, c.3802G>T, c.3811G>T, c.3847C>T, c.3856dupA, c.3875T>C, c.3881T>C, c.3896T>C, c.3905T>C, c.3907A>T, c.3989+1G>A, c.3990-2A>G, c.3990-1G>C, c.3993C>G, c.4015C>T, c.4034G>A, c.4036C>T, c.4075dupA, c.4084T>C, c.4102\_4103dupGC, c.4113\_4114insCA, c.4138dupA, c.4157C>G, c.4164G>A, c.4185G>C, c.4186-1G>A, c.4213C>T, c.4318C>T, c.4324\_4325dupAT, c.4353+1G>A, c.4353+1G>T, c.4353+2T>C, c.4357\_4358dupCA, c.4357C>T, c.4375G>T, c.4393C>T, c.4441A>T, c.4477dupC, c.4480C>T, c.4507G>T, c.4533+1G>A, c.4534-1G>A, c.4593G>A, c.4601G>A, c.4602G>A, c.4644+1G>A, c.4730C>G, c.4753G>T, c.4783C>T, c.4795C>T, c.4850+1G>A, c.4850+2T>A, c.4851-2A>T, c.4851-2A>G, c.4853G>A, c.4854G>A, c.5029C>T, c.5050G>A, c.5050+1G>A, c.5094dupG, c.5101C>T, c.5122C>T, c.5136G>A, c.5164\_5165insC, c.5178\_5179dupCT, c.5205dupT, c.5211-1G>C, c.5216T>G, c.5225T>A, c.5234T>C, c.5245A>T, c.5297C>G, c.5300+1G>T, c.5355G>A, c.5402A>C, c.5418C>G, c.5428C>T, c.5434G>C, c.5435A>G, c.5444T>C, c.5453dupT, c.5458C>T, c.5534+1G>A, c.5534+1G>T, c.5539G>T, c.5548dupA, c.5607+1G>A, c.5665+1G>T, c.5666-2A>C, c.5668A>T, c.5676dupT, c.5706C>A, c.5706C>G, c.5709G>A, c.5752dupA, c.5782C>T, c.5791C>T, c.5833C>T, c.5895-2A>G, c.5898G>A, c.5908G>T, c.5968C>T, c.5981G>A, c.5982G>A, c.6018\_6019insG, c.6041dupA, c.6051T>A, c.6070C>T, c.6079C>T, c.6103+1G>A, c.6104-2A>T, c.6148C>T, c.6157C>T, c.6196G>T, c.6199C>T, c.6292C>T, c.6316A>T, c.6322G>A, c.6322G>T, c.6397C>T, c.6473C>G, c.6492\_6493dupTG, c.6520A>T, c.6526G>T, c.6620dupG, c.6745dupG, c.6757G>T, c.6766C>T, c.6775\_6775+1insGT, c.6775+1G>A, c.6850C>T, c.6884C>G, c.6888\_6889insT, c.6937-2A>G, c.6937-1G>C, c.6955C>T, c.6991A>T, c.7075C>T, c.7132G>T, c.7141\_7142insGT, c.7160C>A, c.7164+1G>A, c.7195C>T, c.7252C>T, c.7276C>T, c.7282C>T, c.7302dupA, c.7367C>G, c.7400T>A, c.7422dupG, c.7425dupT, c.7441C>T, c.7447G>T, c.7451dupT, c.7454dupT, c.7456C>T, c.7485\_7486dupGC, c.7636G>T, c.7824T>A, c.7879C>T, c.7884\_7885dupTA, c.7891C>T, c.7933G>T, c.7957C>T, c.8016G>A, c.8055G>A, c.8077-1G>A, c.8093C>A, c.8356G>T, c.8682\_8683insT, c.8737dupC, c.8956\_8957insA, c.469C>T

<b>Disease: Apert syndrome</b>	<b>Gene: FGFR2</b>	<b>Gene OMIM Number: 176943</b>	<b>NM ID: NM_000141.4</b>	<b>NO.of Variants:2</b>	<b>Phenotype OMIM Number: 101200</b>
<b>Variant: c.940-2A&gt;G, c.758C&gt;G</b>					