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Next Generation Sequencing

Ion AmpliSeq™ GBMv2Pool123_150415v2 (Neuropanel)

Deze assay omvat de coderende sequenties van ATRX (97% CDS), TP53 (98% CDS), PTEN (94% CDS), CIC (76% CDS) en FUBP1 (97% CDS) en hot-spot mutaties in IDH1, IDH2, BRAF, H3F3A en TERT.

Een gemiddelde coverage van >200 reads geeft een sensitiviteit van 10% mutant allel.

Gen	Referentie sequentie	Geanalyseerde exonen (codons)
ATRX	NM_000489	1 (1-7), 2(8-45), 3 (46-63), 4 (64-81), 5 (82-124), 6 (125-162), 7 (163-198), 8 (201-221), 9 (222-289, 299-1097, 1113-1246), 10 (1247-1270), 11 (1276-1315), 12 (1316-1374), 13 (1375-1405), 14 (1406-1439), 15 (1440-1516), 16 (1520-1567), 17 (1568-1603), 18 (1604-1652), 19 (1653-1712), 20 (1713-1758), 21 (1759-1816), 22 (1817-1856), 23 (1857-1899), 24 (1900-1929), 25 (1930-1986), 26 (1987-2037), 27 (2038-2073), 28 (2074-2109), 29 (2110-2114, 2139-2168), 30 (2169-2233), 31 (2234-2283), 32 (2284-2325), 33 (2326-2357), 34 (2358-2400), 35 (2401-2493)
BRAF	NM_004333	11 (439-478), 15 (582-613)
CIC	NM_015125	1 (4-23), 2 (24-68), 3 (74-144), 4 (152-194), 5 (195-255), 6 (256-269), 7 (312-378), 8 (387-441), 9 (455-488), 10 (489-555, 558-758, 784-826, 827-871, 877-900), 11 (901-962), 12 (963-1003), 13 (1004-1059), 14 (1060-1094, 1123-1157), 15 (1158-1178, 1185-1218), 16 (1266-1271, 1345-1347), 17(1348-1358, 1394-1399), 18 (1400-1443), 19 (1444-1487), 20 (1488-1530)
FUBP1	NM_003902	1 (1-40), 2 (48-71), 3 (72-84), 4 (85-97), 5 (98-108), 6 (116-139), 7 (140-158), 8 (159-212), 9 (213-245), 10 (246-279), 11 (280-314), 12 (315-347), 13 (348-395), 14 (396-448), 15 (449-499), 16 (500-526), 17 (527-569), 18 (570-594), 19 (595-642), 20 (643-645)
H3F3A	NM_002107	2 (1-35)
IDH1	NM_005896	4 (105-138)
IDH2	NM_002168	4 (127-155, 159-178)
PTEN	NM_000314	1 (1-27), 2 (28-55), 3 (56-70), 4 (71-85), 5 (86-164), 6 (165-209), 7 (213-267), 8 (268-296, 319-342), 9 (343-404)
TERT	NM_198253	(c.-196 – c.-64)
TP53	NM_000546	2 (1-25), 4 (33-125), 5 (126-187), 6 (188-224), 7 (225-261), 8 (262-307), 9 (308-331), 10 (332-367), 11 (368-394)
Chromosoom	**CNV detectie**	**SNP**
Chr1	LOH, 1p19q	rs8128, rs169957, rs1052230, rs1052231, rs155287, rs169885, rs157208, rs8888, rs7663, rs438228, rs9557, rs7903, rs6424444, rs7504, rs5680, rs191142, rs6604120, rs7686, rs185578, rs12071930, rs12071931, rs1711347, rs2991347, rs2991346, rs504816, rs87061, rs12754569, rs54396, rs54397, rs106075, rs347003, rs1132, rs192990, rs10746395, rs11811946, rs169053, rs178583, rs189882, rs4082255, rs7315, rs7374, rs28734113, rs2274257, rs3736909
Chr19	LOH, 1p19q	rs120959, rs11671746, rs8355, rs11665, rs12852, rs3817, rs72087, rs405152, rs7028, rs33841, rs7283, rs166539, rs3814, rs1291, rs75660813, rs193040, rs10415253, rs2542297, rs12608598, rs2645156, rs10217, rs1135766,

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		rs1052023, rs11573, rs10113, rs2271095, rs6612, rs10448, rs640000, rs8103567, rs17626, rs17628, rs4476273, rs45446093
Chr7	Allelische imbalans, EGFR amp	rs7456393, rs2028209, rs669641, rs3113265, rs1479842, rs2906649, rs6968446, rs7780487, rs2072453, rs730437, rs2075110, rs6950826, rs2345943, rs4724083, rs397970716, rs7796391, rs190, rs39747, rs38852, rs38855, rs38858, rs7795088, rs1399004, rs2523034, rs6978354, rs1208525, rs2402459, rs712735, rs7803170, rs2368058, rs10276566, rs1860793, rs1988377, rs1988378, rs983613, rs2037588, rs2037589, rs7796909, rs7796928, rs1627815, rs10950076, rs11980024, rs4724913, rs2057932, rs853035, rs7791938, rs7777705, rs7800039, rs6957957, rs6975598, rs6955508, rs10237641, rs1167796, rs12537100, rs1263572, rs1525469, rs2390409, rs2681052, rs631085, rs7801597, rs8191992, rs1167795, rs1554497, rs2058447, rs35862642, rs622514, rs185060587, rs17290169, rs7785169, rs80111598, rs13224673
Chr9	LOH, CDKN2A del	rs911602, rs2518720, rs2811710, rs11515, rs10757266, rs589627, rs10120675, rs7869004, rs10757260, rs12335941, rs6475591, rs295264, rs623871, rs321209, rs7867456, rs7046607, rs7046737, rs7864960, rs12708258, rs6475555, rs10115049, rs634537, rs10122243, rs10738616, rs10965333, rs12237826, rs620511, rs12550896, rs182316209, rs1112389, rs528389, rs2595070, rs4145036, rs447053, rs2381609, rs10960529, rs2492624, rs277595, rs6476915, rs7030705, rs7859262, rs870271, rs10811780, rs1555645, rs7857441, rs1533040, rs4740620, rs111497991
Chr10	LOH	rs7099721, rs17113560, rs2814337, rs3011697, rs12765161, rs2311115, rs4933420, rs2761331, rs6480308, rs881501, rs1903858, rs1234224, rs1236816, rs1234213, rs1234214, rs2735343, rs2248293, rs555895, rs532678, rs701848, rs1616888, rs12254170, rs1148210, rs2170133, rs4919204, rs4919205, rs4919206, rs1322331, rs1018720, rs178448, rs1912333, rs3008363, rs7910169, rs2150599, rs4934517, rs4935058, rs7080801, rs7084402, rs2921446, rs721696, rs1149732, rs10788380, rs11201538, rs2785071, rs760177, rs11202662, rs670472, rs558443, rs10749594, rs1926203, rs1274394, rs12774602, rs1070048, rs10785931, rs1514464, rs1856605, rs1969922, rs2354355, rs4244972, rs4934324, rs532102, rs6586081, rs6586170, rs79218317, rs10749586, rs11259405, rs12219242, rs1890628, rs10887862, rs1572203, rs1780133, rs1780134, rs7913758, rs1058801, rs12360380, rs5786835, rs111879418, rs34201179
Chr12	Allelische imbalans	rs238526, rs3851626, rs457487, rs1874560, rs167510, rs1689582, rs1201644, rs2075432, rs1795845, rs1037435, rs1376994, rs2910104, rs7489231, rs747528, rs7967063, rs1381884, rs4019550, rs4391872, rs2398458, rs1705254, rs315131, rs6560927, rs7964704, rs7972746, rs708893, rs838926, rs10841390, rs7978563, rs559759, rs10772149, rs7975075, rs688517, rs1800164, rs702809, rs702810, rs3858577, rs10877124, rs10877125, rs3907924, rs3847698, rs10784009, rs10877895, rs462010, rs466735, rs7135979, rs10878695, rs6581884, rs2367552, rs2367551, rs4761419, rs6539739, rs1038314, rs1492912, rs2058561, rs11608544, rs12336, rs2193042, rs2651975, rs2651976, rs2711702, rs700480, rs7975029, rs8853, rs11177550, rs6487852, rs1905438, rs75356576, rs79516584, rs2873087