

Supplementary Figure 1. SNP-array, qPCR and MAQ assay results putative *de novo* CNV

of article entitled:

Copy Number Variations in 375 patients with oesophageal atresia and/or tracheoesophageal fistula.

Authors and affiliations

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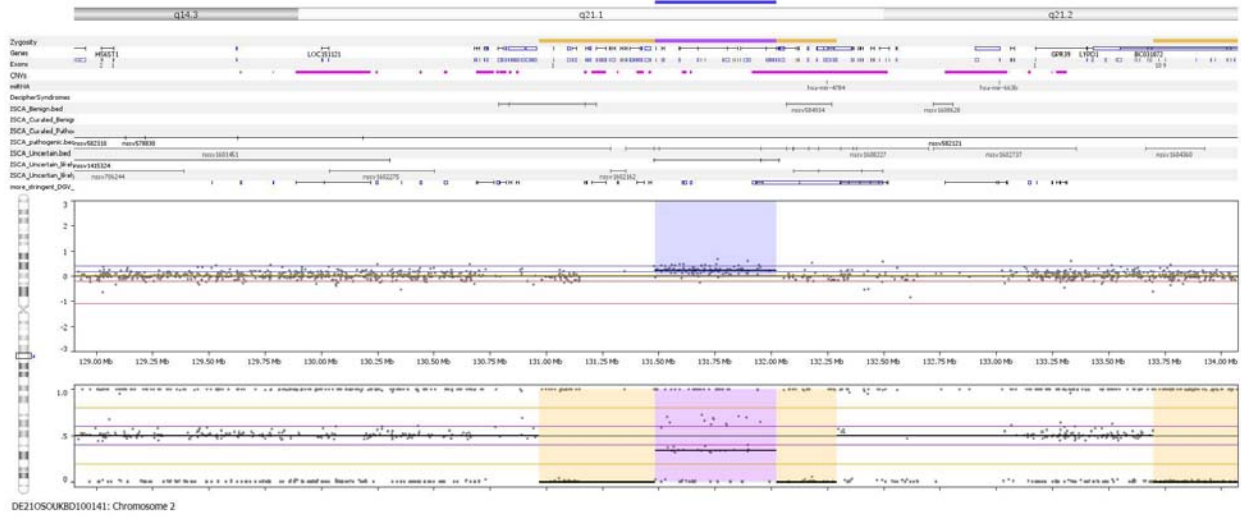
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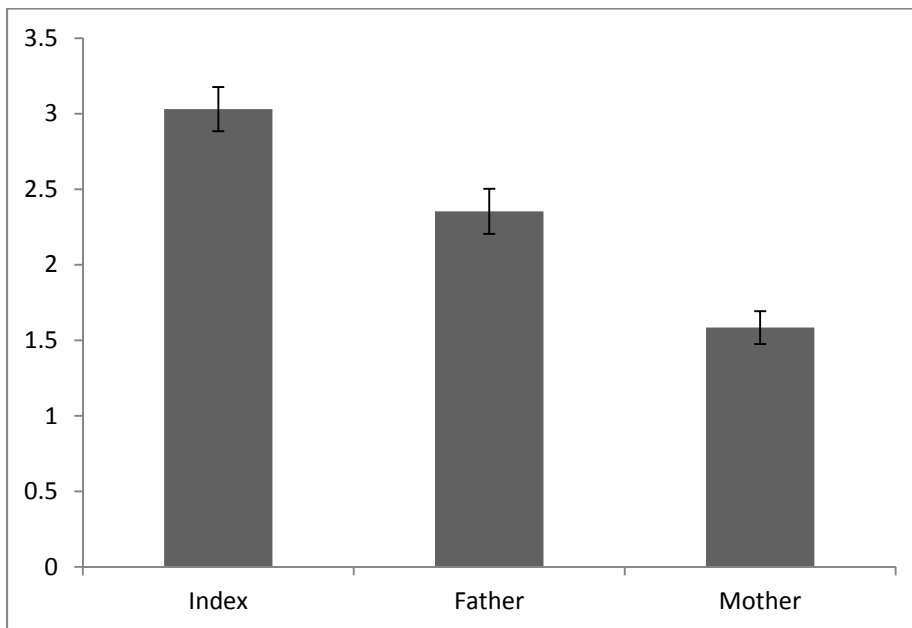
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DE21OSOUKBD100141; Gain 2q21.1 (Chr2:131349899-13191166)



	Forward primer	Reverse primer	Size	Chromosome	Start	Stop
QPCR_1_2q21.1	CAACCTGTGACAAAGCCAGC	TGCAGGGTGTCTTGCTGATG	119	Chr2	131486688	131486806
QPCR_2_2q21.1	AGAGGAAGGACAGCGTGTC	TGACTGTGAGAGCAACTGGG	131	Chr2	131688450	131688580
QPCR_3_2q21.1	GACTGCATCAACATCCGCAC	GGGAAGGAAGAGGACCAAGC	115	Chr2	131883442	131883556

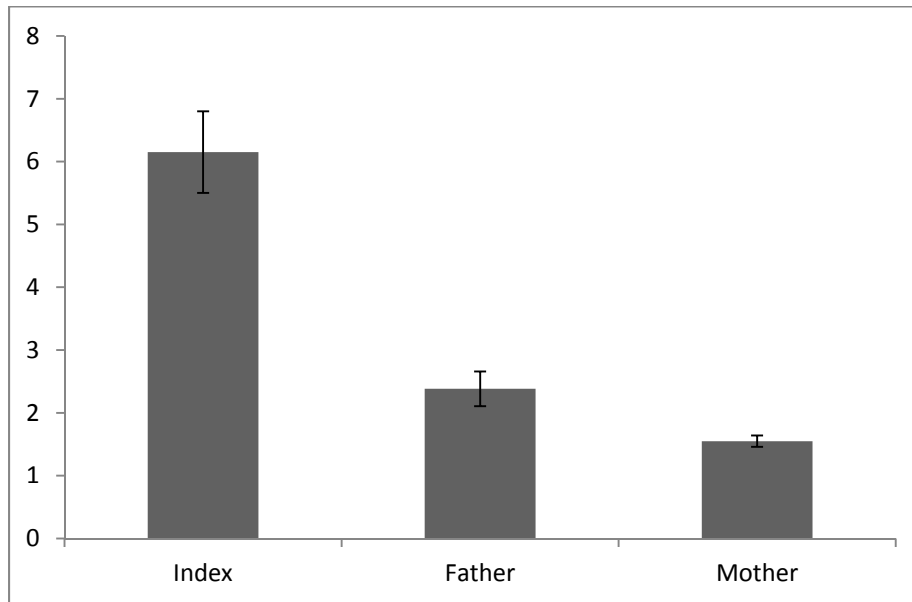


qPCR cutoff for duplication 2.5 and at least two out of three primer pairs matching the criteria.

DE12OSOUKBD100206; Gain 3p26.1(Chr3:8519329-8551649)

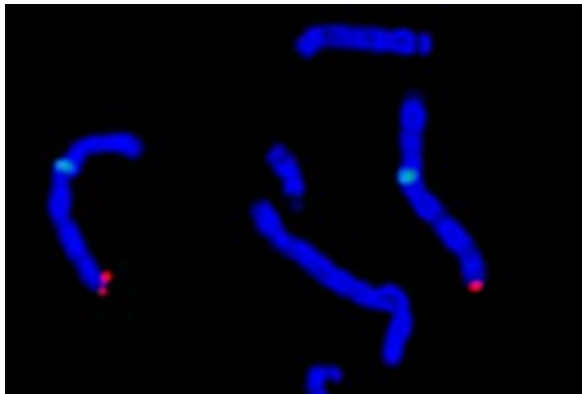
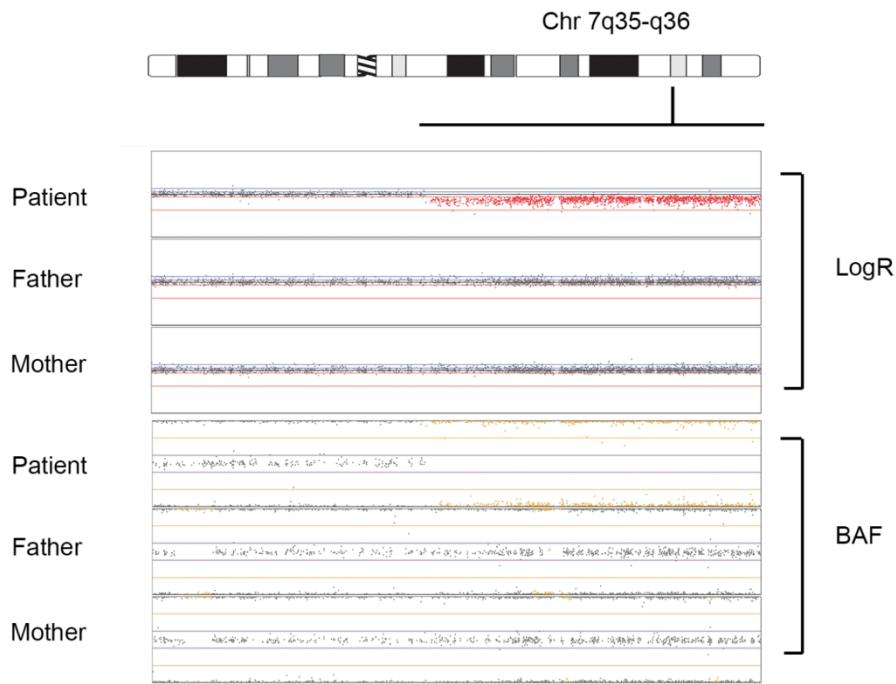


Amplicon	Forward primer	Reverse primer	Size	Chromosome	Start	Stop
QPCR_1_3p26.1	TGACAACGACATCTGCAATCG	TCCAGGGCCACAGTTAACAC	136	Chr3	8523922	8524057
QPCR_2_3p26.1	AGGTGTCCGGTTAAGTCTCTG	TGTAGGCAGAGAGGGGATCC	139	Chr3	8540503	8540641
QPCR_3_3p26.1	GGTGGCTAAGGACCTCAACC	AGTTTCCGCGAACAGATGGG	109	Chr3	8543633	8543741

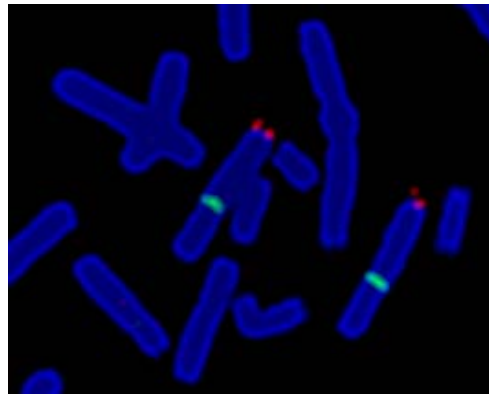


qPCR cutoff for duplication 2.5 and at least two out of three primer pairs matching the criteria.

SKZ_2111; loss 7q35q36 (chr7:143,839,360-159,138,663)



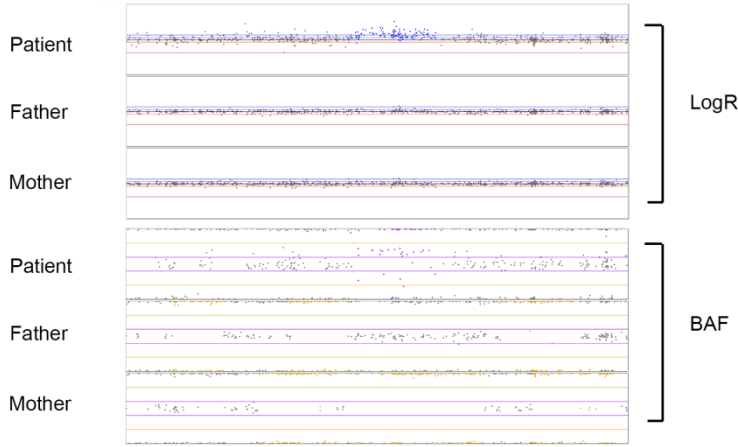
BAC clone 7qter (red+ BAC clone RPI-3K23 (green) in father



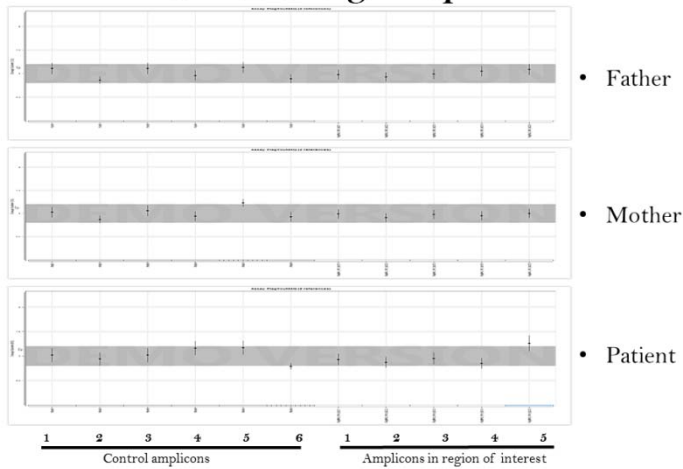
BAC clone 7qter (red+ BAC clone RPI-3K23 (green) in mother

The 7q35q36 loss is not the result of a balanced translocation in one of the parents

SKZ_1810; Gain 8p22 (chr8:17625479-17813225)

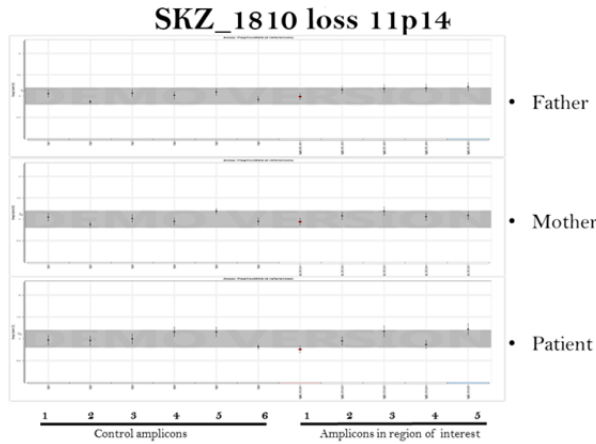
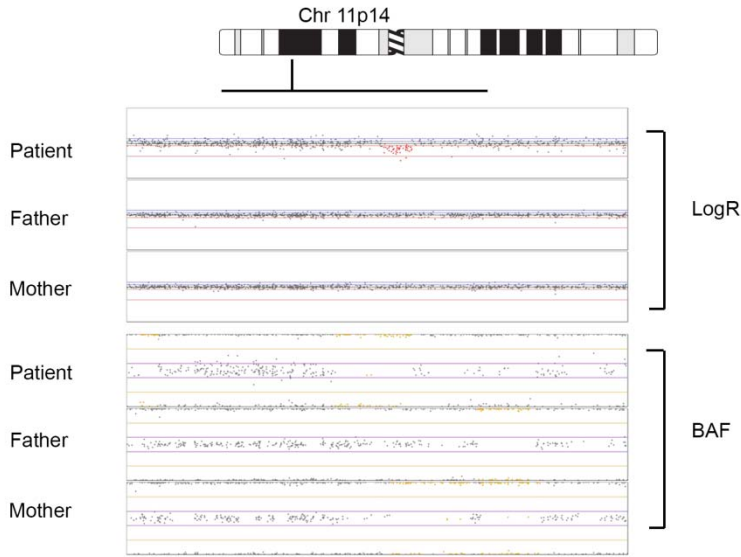


SKZ_1810 gain 8p22



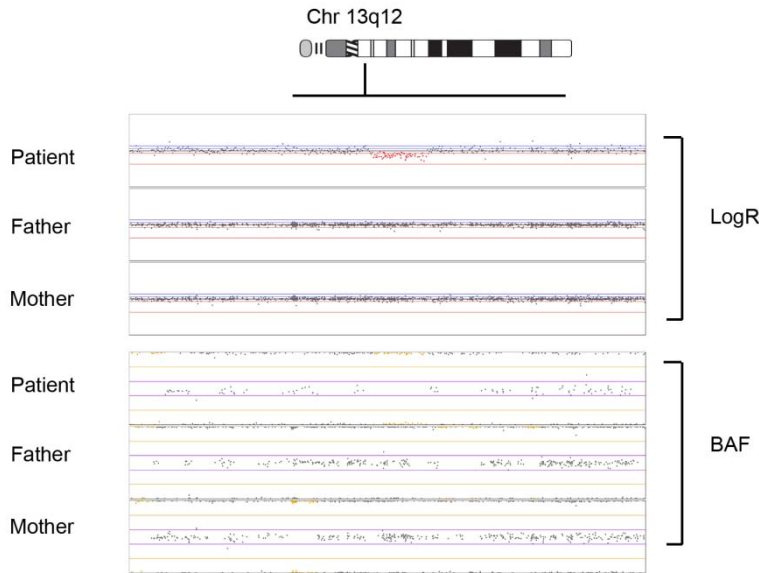
Amplicon	Forward primer	Reverse primer	Size	Chr	Start	Stop
FlagMAQ_1810_8p22.1	5'AGCGGATAACAATTCACACAGGCTCTCAACTCCAAATCTCAACC-3'	5'-GTTTCTTGATGTGTACGATTAGATTAAGGCAGA-3'	245	chr8	17545893	17546107
FlagMAQ_1810_8p22.2	5'AGCGGATAACAATTCACACAGGAGCCATGTGCTGCAAACC-3'	5'-GTTTCTTTGAAGCCTCAAGTGTGAAAGC-3'	169	chr8	17600090	17600228
FlagMAQ_1810_8p22.3	5'AGCGGATAACAATTCACACAGGCCAAGTGAACCTCCAAGC-3'	5'-GTTTCTTGATGAAGTGTCTCCACGAACC-3'	214	chr8	17669963	17670146
FlagMAQ_1810_8p22.4	5'AGCGGATAACAATTCACACAGGCACAACCTGCACCAAAAGCTA-3'	5'-GTTTCTTTGGCAGGCACAATACAGG-3'	295	chr8	17754984	17755248
FlagMAQ_1810_8p22.5	5'AGCGGATAACAATTCACACAGGAGCTAACATGGCAACAGAATAATG-3'	5'-GTTTCTTGGAAAGAGAATGTCCAAATATGC-3'	180	chr8	17823952	17824101

SKZ_1810; loss 11p14.3 (chr11:21853276-22016176)

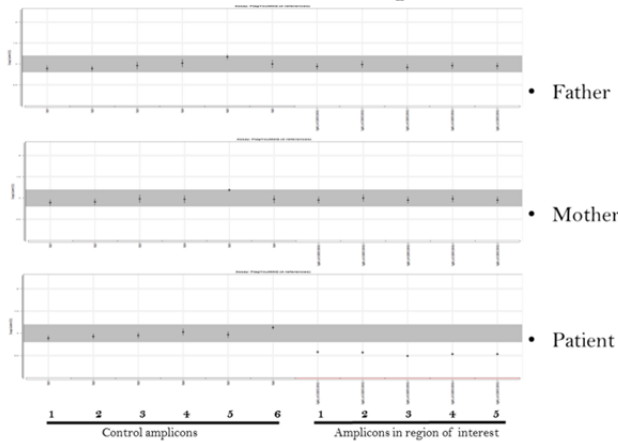


Amplicon	Forward primer	Reverse primer	Size	Chr	Start	Stop
FlagMAQ_1810_11p14.1	5'AGCGGATAACAATTTACACAGGAGGCCTACGAGGGAATGC-3'	5'-GTTTCTTGGGCAAACACCAACAGGA-3'	113	chr11	21795830	21795912
FlagMAQ_1810_11p14.2	5'AGCGGATAACAATTTACACAGGTGAGTATTCACAGGAATAAAGTGG-3'	5-GTTTCTTGGTGCAGGCTTTGAGTCC-3'	197	chr11	21849190	21849356
FlagMAQ_1810_11p14.3	5'AGCGGATAACAATTTACACAGGTGTTGAGCAACCA AGAGAAATTAAG-3'	5-GTTTCTCAACAACATAAGTGTCCACTGA-3'	184	chr11	21918986	21919139
FlagMAQ_1810_11p14.4	5'AGCGGATAACAATTTACACAGGAGTGGAGTTATTAAGGCTGAGGA-3'	5-GTTTCTTTGTGGCAGGGAGTGAATG-3'	177	chr11	21996140	21996286
FlagMAQ_1810_11p14.5	5'AGCGGATAACAATTTACACAGGCAGAGCCTCACTACCCACAGA-3'	5-GTTTCTTTTAATATTCGCTCTCAATAATGTCC-3'	136	chr11	22067921	22068026

SKZ_1662; loss 13q12.11 (chr13:22688792-22981935)

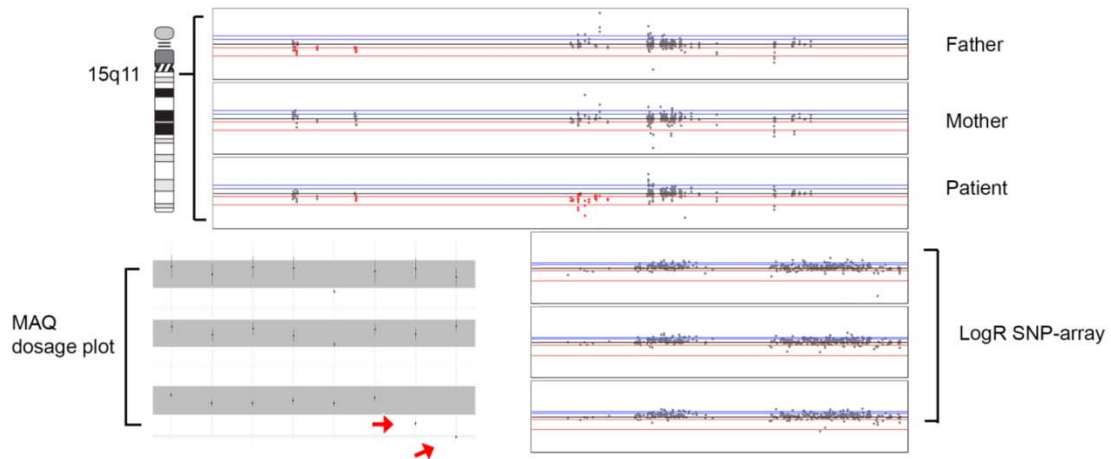


SKZ_1662 loss 13q12.11



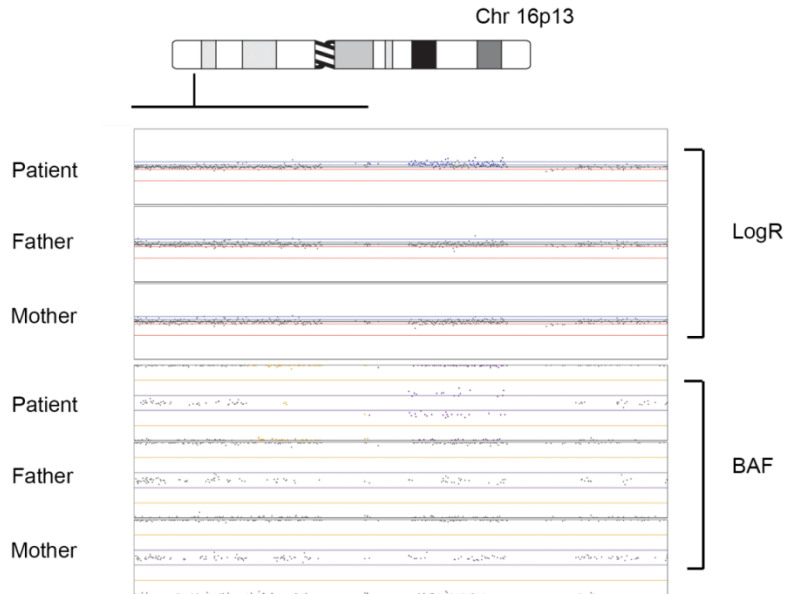
Amplicon	Forward primer	Reverse primer	Size	Chr.	Start	Stop
FlagMAQ_chr13:22688792-22981935.1	5'AGCGGATAACAATTTACACAGGGGTGTAGTTTGCTTAATTCACAGG-3'	5'-GTTTCTTAAAGATACCATCGAAACCACTGA-3'	339	chr13	22696898	22697206
FlagMAQ_chr13:22688792-22981935.2	5'AGCGGATAACAATTTACACAGGAGGACGAGGAAGGCAGA-3'	5'-GTTTCTTGAAAGGACAGGCTGGAAATG-3'	218	chr13	22761030	22761217
FlagMAQ_chr13:22688792-22981935.3	5' AGCGGATAACAATTTACACAGGCTTGTTTCGGGAGTCATAAGTCA-3'	5'-GTTTCTTCCTTTCAATTGTCAACAGCTA-3'	168	chr13	22817621	22817758
FlagMAQ_chr13:22688792-22981935.4	5'AGCGGATAACAATTTACACAGGTGCTTATCACTGAACCACAACC-3'	5'-GTTTCTTAATCACAAAGGAGCTAGAGGAATG-3'	237	chr13	22897279	22897485
FlagMAQ_chr13:22688792-22981935.5	5'AGCGGATAACAATTTACACAGGGACTTCCAACACCACAACTAATG-3'	5'-GTTTCTTCTAGACACAGCTACCAATGC-3'	287	chr13	22965582	22965838

SKZ_0092; loss 15q11

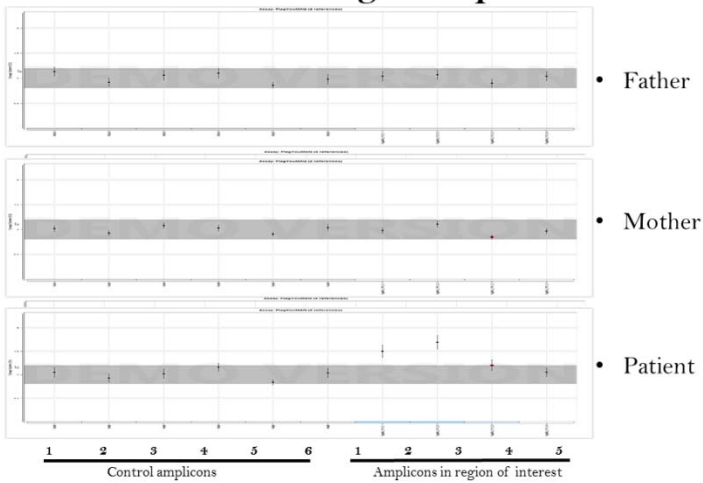


One polymorphism, in the 15q11 region was detected with WES-CN. In the upper panel red dots indicate loss, two MAQ assay probes (red arrow) inside the deleted region, confirm this loss. SNP-array resolution was not enough to call this loss. This is not one of the 167 rare CNVs.

SKZ_2111; Gain 16p13.3(chr16:14,985,615-17,000,304)



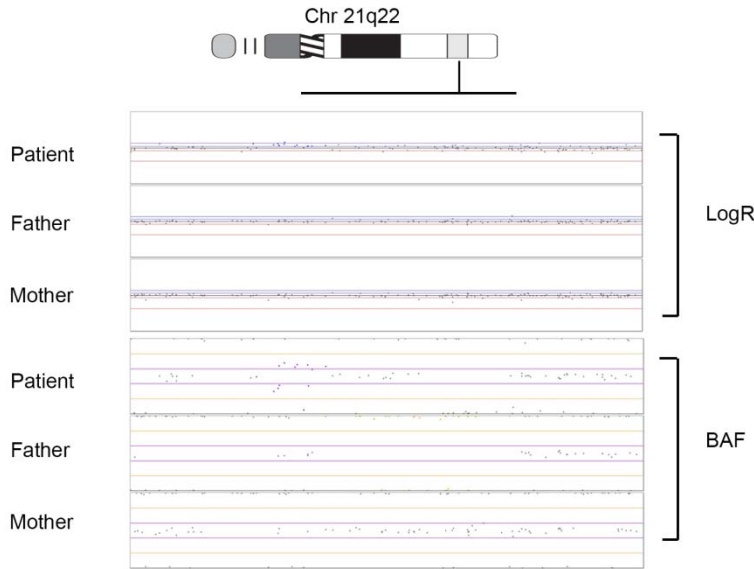
SKZ_2111 gain 16p13



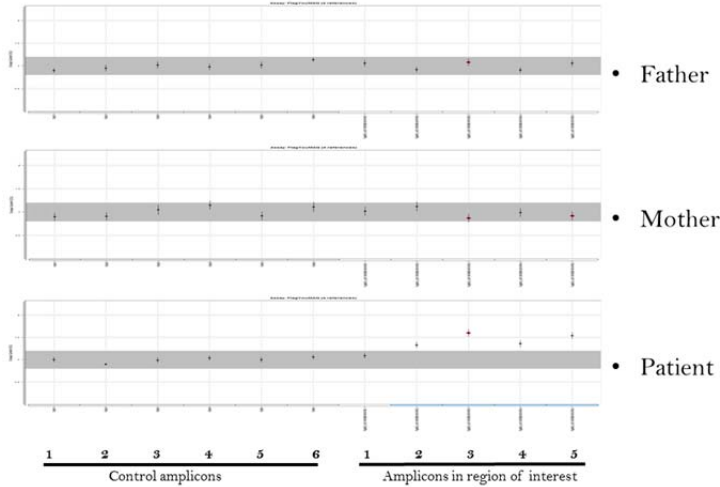
Gain in HG19:

Amplicon	Forward primer	Reverse primer	Size	Chr	Start	Stop
FlagMAQ_2111_2.1	5'AGCGGATAACAATTTACACAGGCAGGGAGACACCATGCAGA-3'	5'-GTTTCTTGGTATGATGAGAGAGGTGAAGGA-3'	173	chr16	15141570	15141712
FlagMAQ_2111_2.2	5'AGCGGATAACAATTTACACAGGGTCGACCCTCCCAAACC-3'	5'-GTTTCTTGGCAATGGAAGATGAAGAGGA-3'	126	chr16	15586155	15586250
FlagMAQ_2111_2.3	5'AGCGGATAACAATTTACACAGGCTGGGTGAAGAGGCCAAAGT-3'	5'-GTTTCTTGGCCACCCTACAGACAGA-3'	134	chr16	16061100	16061203
FlagMAQ_2111_2.4	5'AGCGGATAACAATTTACACAGGTGGATGTGAGAAATGCCAAGT-3'	5'-GTTTCTTGGAGGATGAGGAGCAAACC-3'	216	chr16	16965277	16965462

SKZ_21q22.2; Gain 21q22.2 (chr21:40,095,493-40,160,135)



SKZ_1307 gain 21q22.2



Amplicon	Forward primer	Reverse primer	Size	Chr.	Start	Stop
FlagMAQ_chr21:40100880-40154748.1	5'AGCGGATAACAATTCACACAGGAACCTCGCCTCTTGAACC-3'	5'-GTTTCTTAATTCCTCATCACATGGGTCA-3'	131	chr21	40102211	40102311
FlagMAQ_chr21:40100880-40154748.2	5'AGCGGATAACAATTCACACAGGCTCCAAATGCACTGAAATG-3'	5'-GTTTCTTAGGTGATAGAATGGGTGCTCTTA-3'	271	chr21	40117025	40117265
FlagMAQ_chr21:40100880-40154748.3	5'AG CGGATAACAATTCACACAGGCAGGCAGACAGCTCACGTT-3'	5'-GTTTCTTAGAGAGGCACAGGCACAGA-3'	100	chr21	40125236	40125305
FlagMAQ_chr21:40100880-40154748.4	5'AGCGGATAACAATTCACACAGGCTGCAAAATACAGTGCAAACTGA-3'	5'-GTTTCTTTCTCCATCACITTTCTTAACTTC-3'	122	chr21	40144161	40144252
FlagMAQ_chr21:40100880-40154748.5	5'AGCGGATAACAATTCACACAGGCAGAGAGCAACAAACAATGC-3'	5'-GTTTCTTGCTGTGGTGATGAGGGCTA-3'	110	chr21	40154178	40154257