

Supplemental_Table_S12. Densitometric analysis of splice defects due to selected *ABCA4* variants. For each fragment, intensity measurement was performed in duplicate (Raw intensity 1 and 2) and corrected for fragment size (corrected intensity 1 and 2). For fragments marked with an asterisk (*), sequence results were not available. Highlighted in red are the fragment sizes that were arbitrarily assessed in the absence of sequenced fragments of the same size. (#) indicates the fragments for which sequencing results were obtained from larger amplicons (Supplemental Fig. S3).

BA_variant	Fragment	Raw intensity_1	Raw intensity_2	Fragment size (bp)	Correction factor	Corrected intensity_1	Corrected intensity_2	%_t1	%_t2	Average_%	Remarks	Sequence result available
BA11_WT	*3	16158,87	16329,87	119	1,00	16158,87	16329,87	1,00	1,00	100,0%		yes#
c.1937+13T>G	*4	13225,00	13467,82	131	1,10	12013,55	12234,13	0,86	0,86	86,0%		yes#
c.1937+13T>G	*3	1905,52	2052,35	119	1,00	1905,52	2052,35	0,14	0,14	14,0%	excluded (intensity <15%)	yes#
						13919,06	14286,48	1,00	1,00			
BA_variant	Fragment	Raw intensity_1	Raw intensity_2	Fragment size (bp)	Correction factor	Corrected intensity_1	Corrected intensity_2	%_t1	%_t2	Average_%	Remarks	Sequence result available
BA12_WT	1	6077,38	6369,86	788	1,00	6077,38	6369,86	0,59	0,60	59,7%		yes
BA12_WT	2	2976,06	3052,77	566	0,72	4143,35	4250,15	0,41	0,40	40,3%		yes
						10220,73	10620,01	1,00	1,00			
c.2382+5G>C	*3	1521,36	1581,60	2073	2,63	578,31	601,21	0,03	0,03	2,9%	excluded (intensity <15%)	no
c.2382+5G>C	*4	539,41	531,70	1181	1,50	359,91	354,77	0,02	0,02	1,8%	excluded (intensity <15%)	no
c.2382+5G>C	1	9758,97	9652,85	788	1,00	9758,97	9652,85	0,48	0,48	47,9%	wild-type band	yes
c.2382+5G>C	2	5153,15	5163,86	566	0,72	7174,36	7189,26	0,35	0,36	35,5%		yes
c.2382+5G>C	5	448,28	396,04	138	0,18	2559,77	2261,46	0,13	0,11	11,9%	excluded (intensity <15%)	yes
						20431,31	20059,54	1,00	1,00			
BA12_WT	1	5136,38	5372,15	788	1,00	5136,38	5372,15	0,45	0,45	45,3%		yes
BA12_WT	2	2829,77	2876,18	566	0,72	3939,68	4004,30	0,35	0,34	34,3%		yes
BA12_WT	*5	391,34	435,75	138	0,18	2234,58	2488,19	0,20	0,21	20,4%	excluded (sequence not validated)	no
						11310,64	11864,64	1,00	1,00			

c.2588G>C	1; 6	5263,50	5353,62	785	1,00	5283,61	5374,08	0,16	0,17	16,7%		yes
c.2588G>C	2; 7	7174,50	7257,62	563	0,71	10041,75	10158,09	0,31	0,32	31,7%		yes
c.2588G>C	*5	2961,79	2808,67	138	0,18	16912,25	16037,91	0,52	0,51	51,6%	excluded (sequence not validated)	no
						32237,61	31570,07	1,00	1,00			