

Figure 3s. (Supplementary Materials)

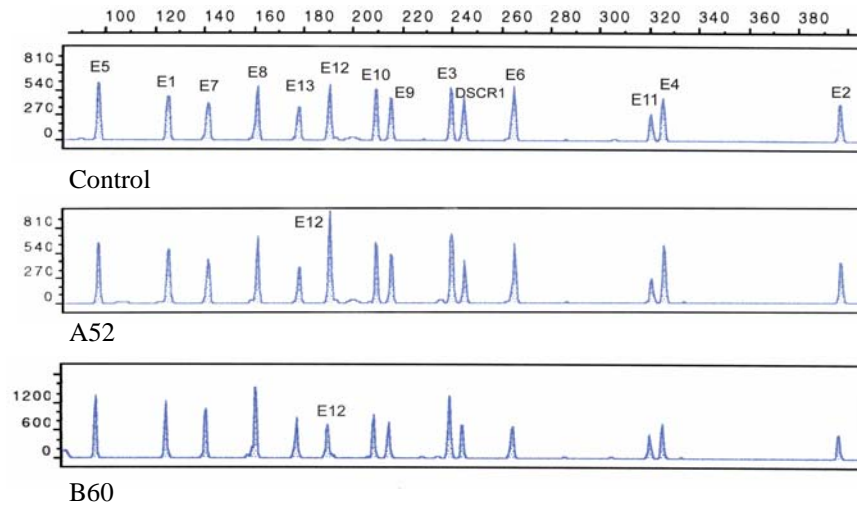


Figure Results semi-

	Family 73						Family 94
<i>SLC7A9</i>	A50	A51	A52	A53	A54	A55	B60
Exon 1	0.99	1.11	0.79	0.94	0.71	0.87	1.03
	1.09	0.88	1.00	0.83	0.83	0.78	1.08
Exon 2	1.04	1.06	1.04	1.24	1.04	1.16	1.03
	0.70	0.76	0.77	0.77	0.75	0.72	1.23
Exon 3	1.27	1.37	1.24	1.56	1.39	1.57	1.19
	0.91	1.13	1.11	1.24	0.95	1.11	1.40
Exon 4	1.26	1.25	1.10	1.36	1.14	1.28	1.07
	0.93	0.82	0.87	0.88	0.78	0.83	1.32
Exon 5	0.79	1.25	0.75	0.92	0.78	0.94	0.84
	1.08	0.83	0.93	0.74	0.72	0.80	1.16
Exon 6	1.06	1.33	1.03	1.24	1.04	1.24	1.07
	0.90	0.88	0.88	0.85	0.74	0.81	1.38
Exon 7	0.91	1.06	0.91	1.10	0.87	1.03	1.10
	0.96	0.88	1.08	0.89	0.70	0.77	1.02
Exon 8	0.91	1.04	0.89	1.21	0.95	1.07	1.11
	1.15	1.00	1.03	0.90	0.57	0.61	1.23
Exon 9	1.02	1.16	0.99	1.16	1.02	1.09	1.18
	0.95	0.98	0.98	0.95	0.94	0.99	1.35
Exon 10	1.13	1.24	0.99	1.11	1.07	1.18	1.15
	0.93	1.01	0.95	1.00	0.95	1.03	1.04
Exon 11	1.05	1.21	1.01	1.07	0.90	1.05	1.07
	1.01	0.91	0.97	0.85	0.97	0.91	1.05
Exon 12	1.59	1.05	1.48	1.06	0.95	1.56	0.63
	1.47	0.91	1.37	0.78	0.89	1.40	0.51
Exon 13	0.90	1.08	0.92	0.89	0.92	0.98	1.09
	1.02	1.01	0.91	0.88	0.93	0.92	0.97

3s.
of two

quantitative multiplex PCR analysis in families 73 and 94. Individuals A52 and B60 have an abnormal pattern (up): A52 have approximately double dose for exon 12 and B60 have half of the dose for exon 12 comparing with the control. The experiment was run with two replicas including all the members of family 73 and B60 from family 94, and the results were confirmed (bottom). The double dose for exon 12 is observed in

A50 (father), A52 (proband) and A55 (brother) (in bold). The segregation in family 94 could not be performed because there was no DNA of other members.