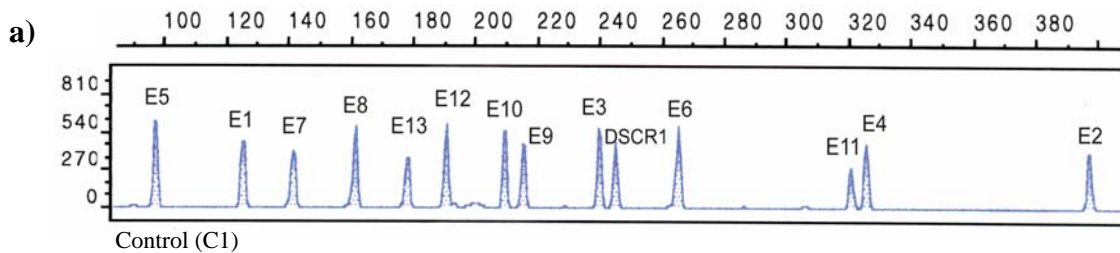


Figure 1s.

Semi-quantitative Multiplex PCR analysis of SLC7A9 in controls. a) Chromatogram of a control (C1). The assignation of areas under peaks was made by GENESCAN™ software. Peak (SLC7A9 exons E1 to E13 and DSCR1-E5) sizes (top ruler in bp) and quantities (left ruler) were determined using the same software and the area of each peak was divided by the external double dose control (exon 5 of DSCR1) (internal to the reaction). We considered that the area of each peak obtained by semiquantitative PCR would be a reliable indicator of the amount of DNA in each exon, so we would see a clear difference in ratios wherever there was a large rearrangement. b) Three unrelated normal controls (C1, C2 and C3) are shown as an example of the reproducibility of the method. The values presented here were obtained as follows: The area below each peak is a measure of the amount of DNA present. These areas were divided by that of the internal double dose control (exon 5 of DSCR1). A mean of the values was obtained for each exon. Last, each value is divided by the mean of its corresponding exon. If all the samples are similar for one particular exon, as is the case with the normal controls, all the values are around 1. When a value is 0 we suspect a complete deletion, when it is 0.5 we suspect a deletion in heterozygosis and when it is 2 or larger we suspect that the sample has a duplication for this exon.



b)

| Exon | Genescan Data | | | Divided by DSCR1-E5 | | | Average | Ratio/Average | | |
|----------|---------------|------|------|---------------------|------|------|--------------|---------------|-------------|-------------|
| | C1 | C2 | C3 | C1 | C2 | C3 | C1 + C2 + C3 | C1 | C2 | C3 |
| 1 | 3448 | 4477 | 4315 | 1.32 | 1.21 | 1.32 | 1.28 | 1.03 | 0.94 | 1.03 |
| 2 | 2834 | 3891 | 4089 | 1.09 | 1.05 | 1.25 | 1.13 | 0.96 | 0.93 | 1.11 |
| 3 | 5247 | 4606 | 6182 | 2.01 | 1.25 | 1.89 | 1.72 | 1.17 | 0.73 | 1.1 |
| 4 | 3566 | 4227 | 4615 | 1.36 | 1.14 | 1.41 | 1.3 | 1.04 | 0.87 | 1.08 |
| 5 | 3555 | 5420 | 4979 | 1.36 | 1.46 | 1.52 | 1.45 | 0.94 | 1.01 | 1.05 |
| 6 | 5059 | 6381 | 6218 | 1.94 | 1.73 | 1.9 | 1.86 | 1.04 | 0.93 | 1.02 |
| 7 | 3046 | 3859 | 3642 | 1.17 | 1.04 | 1.11 | 1.11 | 1.06 | 0.94 | 1.00 |
| 8 | 3973 | 4910 | 4565 | 1.52 | 1.33 | 1.39 | 1.41 | 1.08 | 0.94 | 0.98 |
| 9 | 2445 | 3677 | 3656 | 0.94 | 0.99 | 1.12 | 1.02 | 0.92 | 0.97 | 1.1 |
| 10 | 2480 | 3842 | 3642 | 0.95 | 1.04 | 1.11 | 1.03 | 0.92 | 1.01 | 1.07 |
| 11 | 2343 | 3085 | 2895 | 0.9 | 0.83 | 0.88 | 0.87 | 1.03 | 0.95 | 1.01 |
| 12 | 3704 | 4969 | 4406 | 1.42 | 1.34 | 1.35 | 1.37 | 1.04 | 0.98 | 0.99 |
| 13 | 2722 | 3686 | 3493 | 1.04 | 1.00 | 1.07 | 1.04 | 1.00 | 0.96 | 1.03 |
| DSCR1-E5 | 2613 | 3700 | 3273 | 1 | 1 | 1 | 1 | 1 | 1 | 1 |