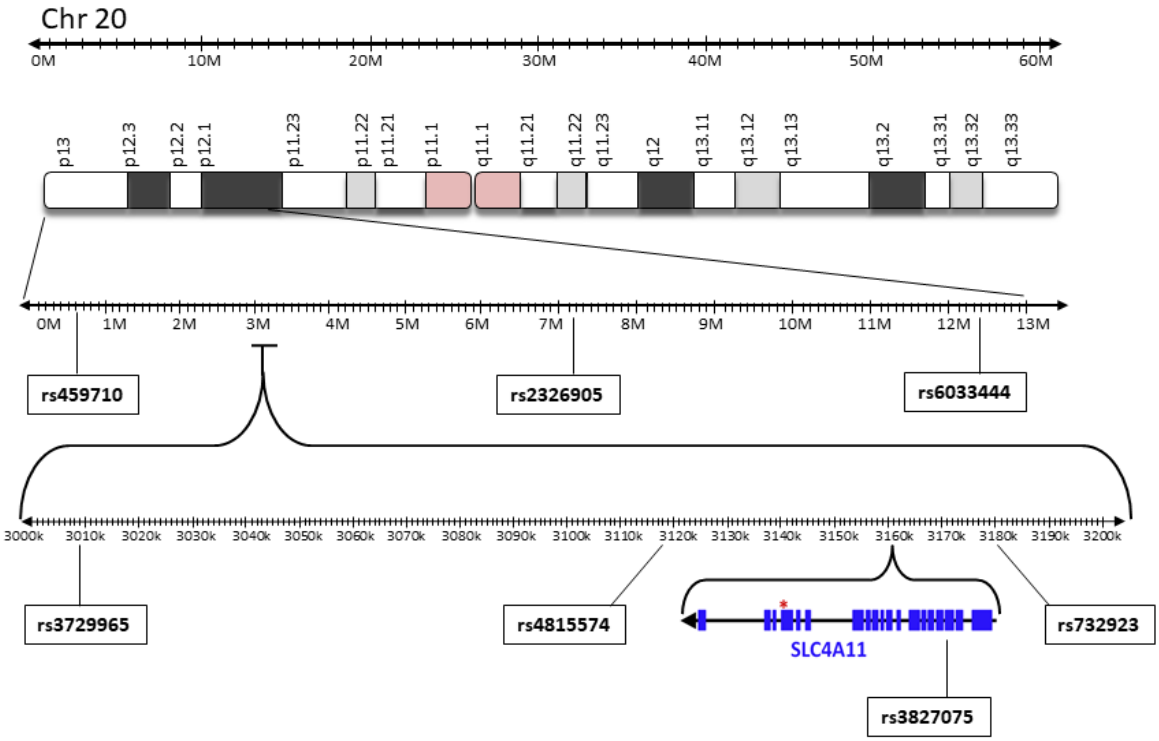
**Supplementary materials for Romero et al., "Clinical features and possible founder mutation of the 8bp duplication mutation in the *SLC4A11* gene causing corneal dystrophy and perceptive deafness in three South American families", *Ophthalmic Genetics,* 2019.**

## Supplementary methods

7 SNPs were selected based on their chromosomal locations covering a region near 12Mb (Supplementary Figure 1). One of the SNP was located inside the SLC4A11 gene (rs3827075) and the other six SNPs surrounding it (three at each side). Other criterion was a minor allele frequency (MAF) of at least 0.2 in Latin-American populations, such as Mexican (Mex) or Peruvian populations, when they were available in dbSNPs (1) or the information of the European population (Ceu) when data for Latin-America was not available. Latin-American population was our principal reference because it is closer to Chilean than other populations (2). One of these SNPs was located within the *SLC4A11* gene (rs3827075), three are located in telomeric direction and the other three in centromeric direction. These SNPs cover a region of approximately 12 Mb of the short arm of the chromosome 20 located in the coordinates indicated in Supplementary Table 1.

We designed methods based on PCR-RFLP under the conditions described in Supplementary Table 1 to genotype the 7 SNPs. The different alleles were defined by electrophoresis in 3% agarose gels. The amplicon sizes and the digestion products length are listed in Supplementary Table 1. The haplotype frequencies in the control population were inferred using SNPstats package (http://bioinfo.iconcologia.net/SNPstats).




**Supplementary Figure 1.** Location of the 7 SNPs (rs732923, rs4815574, rs3827075, rs3729965, rs459710, rs2326905 and rs6033444) used to evaluate the founder mutation on Chromosome 20. These 7 SNPs cover a 12 Mb region near. One of the SNP was located inside the *SLC4A11* gene (rs3827075) and the other six SNPs surrounding it (three at each side). The red Asterisk indicates the location of the c.2233_2240dupTATGACAC duplication mutation in *SLC4A11* gene.

## References

1. Sherry ST, Ward MH, Kholodov M, Baker J, Phan L, Smigielski EM, Sirotkin K. dbSNP: the NCBI database of genetic variation. Nucleic acids research. 2001;29(1):308-11.

2. Adhikari K, Mendoza-Revilla J, Chacon-Duque JC, Fuentes-Guajardo M, Ruiz-Linares A. Admixture in Latin America. Curr Opin Genet Dev. 2016;41:106-14.

**Supplementary Table 1.** Summary of the primers and PCR conditions used for detecting the mutation in all 19 exons and to construct haplotypes. The primers and PCR conditions used for detecting the mutation in all 19 exons includes sequence primers, annealing temperature and amplicon sizes. The primers and PCR conditions used to construct haplotypes includes sequence primers, annealing temperature, amplicon sizes, restriction enzymes, and sizes of digestion products for SNPs and coordinates of each SNP.

| DNA fragments or SNPs | Primers sequences 5´to 3´ | Annealing Temp (ºC) | PCR product length (bp) | Restriction enzyme | Allele and digestion products length (bp) | Coordinate of SNP location (chr20) |
|---|---|---|---|---|---|---|
| Screening of mutation by PCR | CCACTCCCCGCTGCACGTG (F)<br>AGGGTCCACGGCTCTTGCCA (R) | 63 | 105 (normal allele)<br>113 (allele with duplication) | ------- | ---------- | ---------- |
| Exon 1 | CCTGCTTCCCTTTCTCCCT (F)<br>AGCACTAGAGTGGCCCAGAT (R) | 61 | 316 | ------- | ---------- | ---------- |
| Exons 2-3 | gGATGGCCTCTCCCACCAC (F)<br>CTCCCTGTTGAGCTGCTCCT (R) | 61 | 577 | ------- | ---------- | ---------- |
| Exons 4-5 | TCCAGGAGCAGCTCAACAG (F)<br>TCTTCTCCCAAGTTGGTTGG (R) | 61 | 677 | ------- | ---------- | ---------- |
| Exon 6 | CAAGGTCGAGGGGGTTCT (F)<br>GTTTCTGACACACCCACAGG (R) | 61 | 382 | ------- | ---------- | ---------- |
| Exons 7-8 | AGCCTGGGTGACAGTGAGAC (F)<br>ACAGCCTTGTTTTTCCCAAT (R) | 62 | 660 | ------- | ---------- | ---------- |
| Exons 9-10 | ACTGATGGTACGTGGCCTCT (F)<br>CGTCCATGCGTAGAAGGAGT (R) | 61 | 567 | ------- | ---------- | ---------- |
| Exons 11-12 | CTCTTCTCTGGGCAGCCATT (F)<br>ACTCAGCTTGAGCCAGTCCT (R) | 61 | 712 | ------- | ---------- | ---------- |
| Exons 13-14 | GAGCCCTTTCTCCCTGAGAT (F)<br>GGTTGTAGCGGAACTTGCTC (R) | 61 | 623 | ------- | ---------- | ---------- |
| Exons 15-16 | GCCTTCTCCCTCATCAGCTC (F)<br>CCGCGAGTGTCACCTCTG (R) | 61 | 666 | ------- | ---------- | ---------- |

| | | | | | | |
|---|---|---|---|---|---|---|
| Exon 17 | CGTGGACCCTGAGGAGTG (F)<br>CCCTCCGGATGTAGTGTGTC (R) | 61 | 420 | ------- | ---------- | ---------- |
| Exons 18 | CTCGATGGCAACCAGCTC (F)<br>CTAGGCAGGACCCCTCCTC (R) | 61 | 398 | ------- | ---------- | ---------- |
| Exons 19 | CAGGAGGGGCTCCAGTCTA (F)<br>ACAGAGCAGTCACCCACACA (R) | 65 | 337 | ------- | ---------- | ---------- |
| rs459710 | AGCAGTAAGTTATGCCCAGTA (F)<br>GACCCAATCTATAGGAGCAG (R) | 58 | 495 | TaqI | C: 289 and 116 bp<br>T: 495 bp | 411,995 |
| rs3729965 | ATCTTGTTGTCACAGGGCTAA (F)<br>TGCTGCCTCTGCTGGCTGCCT (R) | 60 | 457 | NIaII | C: 255, 171 and 31 bp<br>T: 255 and 202 bp | 3,013,547 |
| rs4815574 | CACCTCATACTGTGGTTGGCT (F)<br>TGACTCTGCTACTTCCATCTGCA (R) | 56 | 143 | PstI | C: 124 and 19 bp<br>T: 143 bp | 3,118,172 |
| rs3827075 | GGTATCTGACAGCAGGTGGA (F)<br>CAGGCAGTGACAGCATCTCA (R) | 58 | 391 | AvaI | G: 77, 111 and 203 bp<br>T: 77 and 314 bp | 3,162,819 |
| rs732923 | GGAGAGGAGTTGGGACAGTG (F)<br>AAGGGCTTGCCTCGCTGCAGA (R) | 60 | 402 | PstI | C: 242,145 and 15 bp<br>G: 387 and 15 bp | 3,179,340 |
| rs2326905 | ACGGCGTTGTGAAACTGCCA (F)<br>CTTGCTGTCATTCGCGCTTA (R) | 58 | 523 | NdeI | T: 356 and 167 bp<br>C: 523 bp | 7,158,137 |
| rs6033444 | GCAATGTGAGCTTTCACTCTG (F)<br>AGTTCCTTGTAGCATCAGGCT (R) | 60 | 507 | TaqI | C: 326 y 181 bp<br>T: 507 bp | 12,459,371 |

**Supplementary Table 2.** Clinical features in the affected and unaffected subjects. The phenotypic features of five patients affected with CDPD and five unaffected subjects of the families are shown. Corneal phenotypes were assessed by slit lamp examination. The following abbreviations are used: years (Y), male (M), female (F), right eye (OD), left eye (OS).

| Patient | Gender | Age (Y) | Hearing loss | Age of onset of Hypoacusia (Y) | Corneal Dystrophy | Age of onset of Dystrophy (Y) | Best corrected Vision | | Corneal Graft Rejection | |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | OD | OS | OD | OS |
| **Family 1** | | | | | | | | | | |
| III-3 | M | 44 | No | - | No | - | 1.0 | 1.0 | - | - |
| III-11 | F | 41 | Yes | 3 | No | - | 1.0 | 1.0 | - | - |
| IV-7 | M | 23 | Yes | 10 | Yes | 1 | 0.3 | 0.2 | Yes | No |
| IV-8 | F | 21 | No | - | No | - | 1.0 | 1.0 | - | - |
| IV-9 | M | 15 | Yes | 12 | Yes | 1 | 0.1 | 0.1 | No | Yes |
| IV-10 | F | 11 | Yes | 8 | Yes | at birth | 0.2 | 0.2 | No | No |
| **Family 2** | | | | | | | | | | |
| IV-5 | F | 40 | Yes | 18 | Yes | at birth | 0.1 | 0.1 | Yes | Yes |
| III-6 | F | 65 | No | - | No | - | 1.0 | 1.0 | - | - |
| **Family 3** | | | | | | | | | | |
| II-2 | M | 28 | No | - | No | - | 1.0 | 1.0 | - | - |
| II-3 | F | 28 | No | - | No | - | 1.0 | 1.0 | - | - |
| III-1 | M | 12 | Yes | 11 | Yes | at birth | 0.2 | 0.2 | No | No |