

## Supplementary Material to:

## Two Single Nucleotide Deletions in the *ABCD1* Gene Causing Distinct Phenotypes of X-Linked Adrenoleukodystrophy

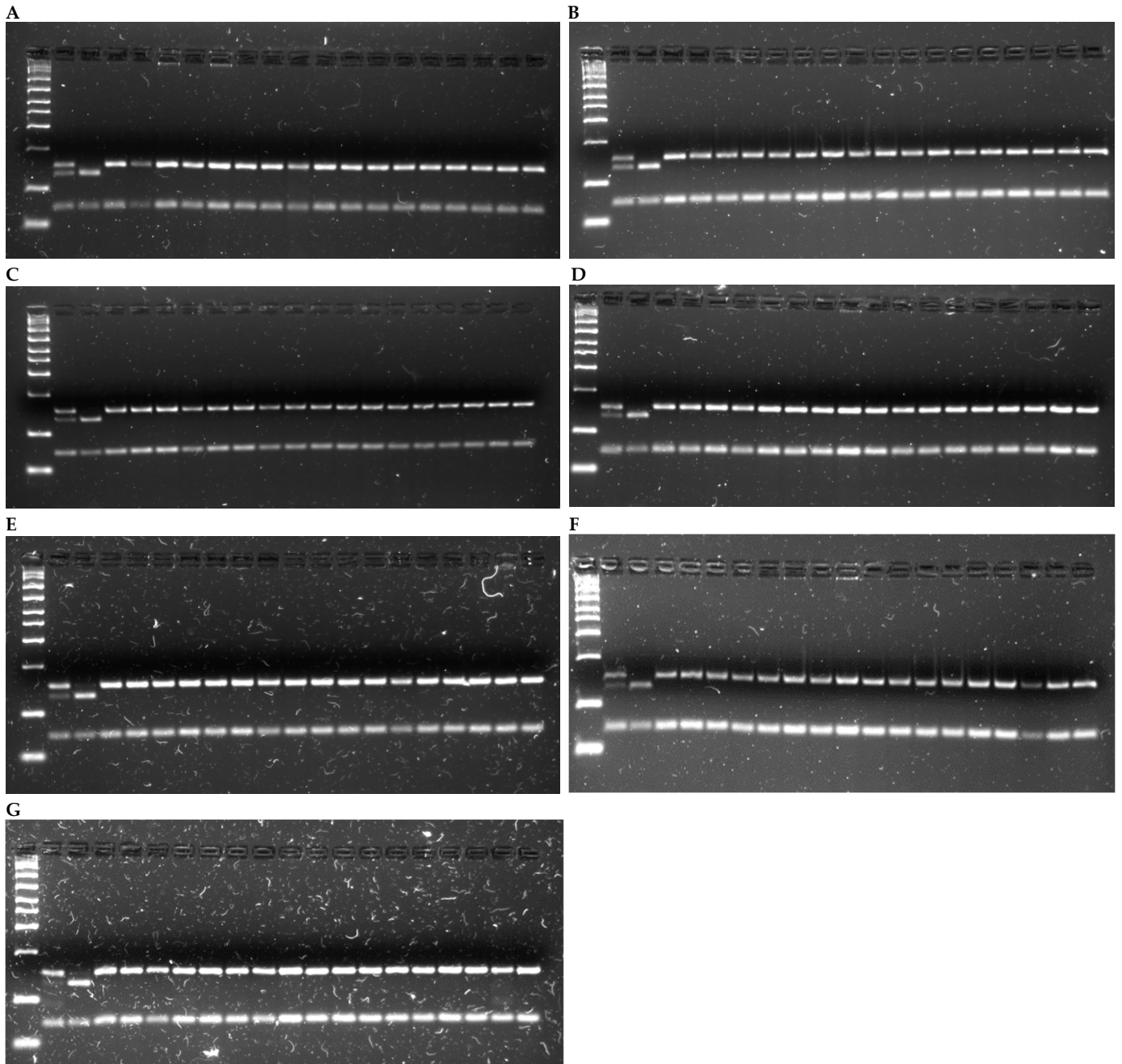
**Supplementary Table S1.** M13-tag primer for Sanger Sequencing of ABCD1 gene.

| Name          | Sequence  |
|---------------|---|
| ABCD1_1F_M13  | <b>CACGACGTTGTAAAACGACCATCGCAAGGTTTCCAGTT</b>   |
| ABCD1_1R_M13  | <b>GGATAACAATTTACACAGGTGGCACTGTTGACGAAGGTA</b>  |
| ABCD1_1AF_M13 | <b>CACGACGTTGTAAAACGACGCACCTTCTGTCTGGTGTAT</b>  |
| ABCD1_1AR_M13 | <b>GGATAACAATTTACACAGGCACACGCGAGTGCATGTAG</b>   |
| ABCD1_1BF_M13 | <b>CACGACGTTGTAAAACGACCGTGGCTGTGACTTCCTACA</b>  |
| ABCD1_1BR_M13 | <b>GGATAACAATTTACACAGGCAGCTCTAAGGCCAGGACAA</b>  |
| ABCD1_2F_M13  | <b>CACGACGTTGTAAAACGACCCACCAATCGTAACCTCTG</b>   |
| ABCD1_2R_M13  | <b>GGATAACAATTTACACAGGCTGTGCCTGGAGAAGTGACA</b>  |
| ABCD1_3F_M13  | <b>CACGACGTTGTAAAACGACGATGTGCTCTGGGTTGGTTT</b>  |
| ABCD1_3R_M13  | <b>GGATAACAATTTACACAGGATAGGGAGGGGACAGCCTTA</b>  |
| ABCD1_4F_M13  | <b>CACGACGTTGTAAAACGACCATCCTTGCCATGCTTCTCT</b>  |
| ABCD1_4R_M13  | <b>GGATAACAATTTACACAGGGCAGCTACTGTCTGGGAAGG</b>  |
| ABCD1_5F_M13  | <b>CACGACGTTGTAAAACGACGACACTGGGGGAAGAGTTCA</b>  |
| ABCD1_5R_M13  | <b>GGATAACAATTTACACAGGTTAGGAGTGTGGGGAGTTGG</b>  |
| ABCD1_6F_M13  | <b>CACGACGTTGTAAAACGACCTCTCAAGGCTGGTCAGGAG</b>  |
| ABCD1_6R_M13  | <b>GGATAACAATTTACACAGGCAGATCCAAAACAGGGCAGT</b>  |
| ABCD1_7F_M13  | <b>CACGACGTTGTAAAACGACCTGCCCTGTTTTGGATCTGT</b>  |
| ABCD1_7R_M13  | <b>GGATAACAATTTACACAGGCCATCTGTGTGGTGTGGTC</b>   |
| ABCD1_8F_M13  | <b>CACGACGTTGTAAAACGACGGCTTGACTCCACCGTATC</b>   |
| ABCD1_8R_M13  | <b>GGATAACAATTTACACAGGGCTCAGGCTCCACTGAGCC</b>   |
| ABCD1_9F_M13  | <b>CACGACGTTGTAAAACGACTGGGTGCTGGTGGAAGTGA</b>   |
| ABCD1_9R_M13  | <b>GGATAACAATTTACACAGGCTGCTGATGACAGCCGCCT</b>   |
| ABCD1_10F_M13 | <b>CACGACGTTGTAAAACGACAGGCGGCTGTCATCAGCA</b>    |
| ABCD1_10R_M13 | <b>GGATAACAATTTACACAGGCGGGAAGGGTTTTCTAGGAGG</b> |

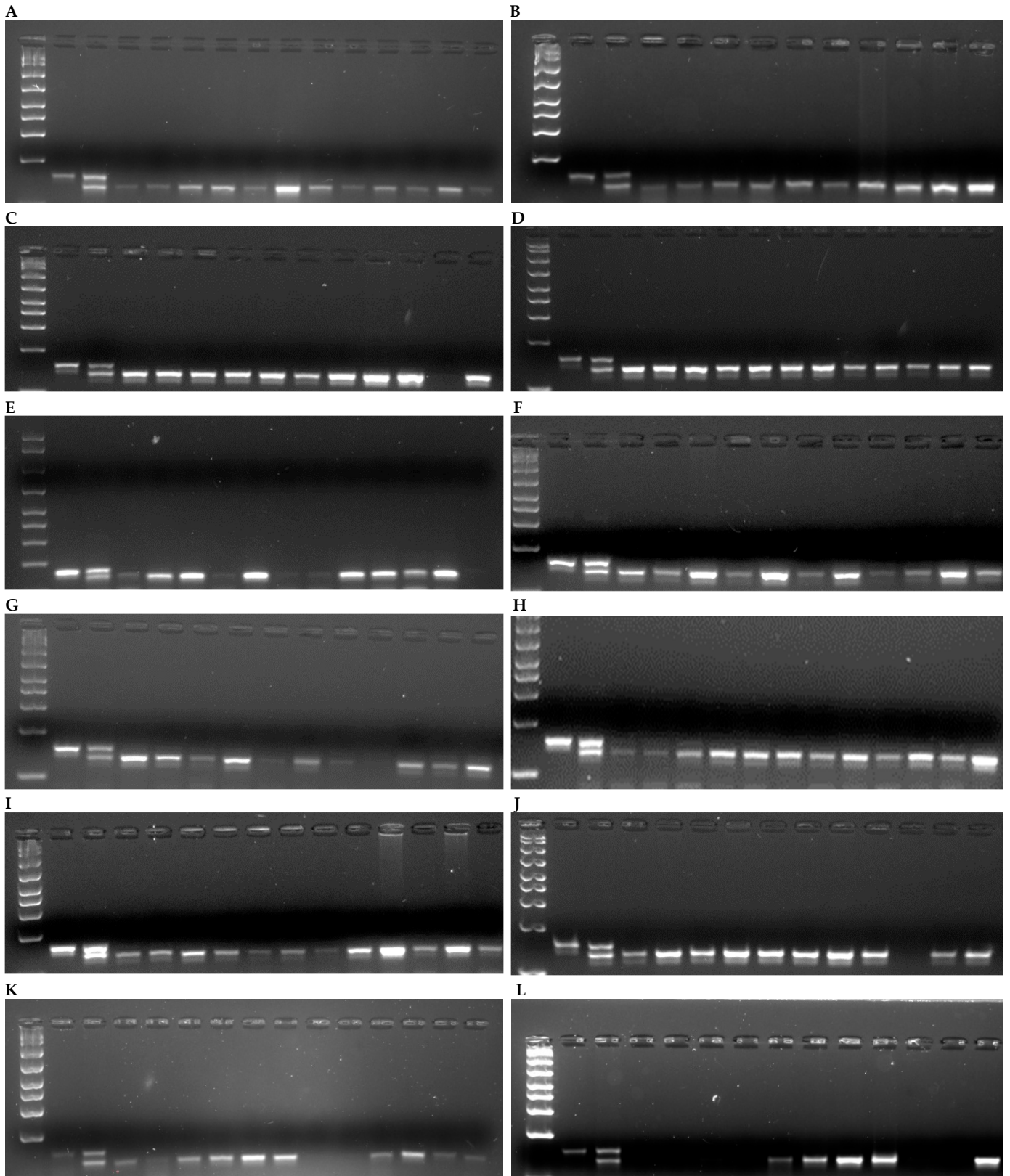
The M13-tag sequence is depicted in bold.

**Supplementary Table S2.** Primer pairs used for RT-qPCR.

| Name                 | Sequence              |
|----------------------|-----------------------|
| ABCD1 exon 1_forward | CCTACTACGGGTCAGCAAC   |
| ABCD1 exon 1_reverse | GAGTAGAGGTGGGCCACAGA  |
| ABCD1 exon 4_forward | GTGCACGAGATGTTCCAGGT  |
| ABCD1 exon 4_reverse | CGGCCTATGGTCCCAGAC    |
| GAPDH_forward        | AAGGTCATCCCTGAGCTGAA  |
| GAPDH_reverse        | TTTGGCAGGTTTTTCTAGACG |



**Figure S1:** Frequency determination of the variant c.253delC in 119 people not suffering from X-ALD. (A) (1) standard, (2) mother (heterozygous), (3) index patient (hemizygous), (4-20) wildtype female. (B) (1) standard, (2) mother (heterozygous), (3) index patient (hemizygous), (4-20) wildtype female. (C) (1) standard, (2) mother (heterozygous), (3) index patient (hemizygous), (4-20) wildtype female. (D) (1) standard, (2) mother (heterozygous), (3) index patient (hemizygous), (4-12) wildtype female, (13-20) wildtype male. (E) (1) standard, (2) mother (heterozygous), (3) index patient (hemizygous), (4-20) wildtype male. (F) (1) standard, (2) mother (heterozygous), (3) index patient (hemizygous), (4-20) wildtype male. (G) (1) standard, (2) mother (heterozygous), (3) index patient (hemizygous), (4-20) wildtype male.



**Figure S2:** Frequency determination of the variant c.1275delA in 119 people not suffering from X-ALD. (A) (1) standard, (2) index patient (hemizygous), (3) sister (heterozygous), (4-15) wildtype male. (B) (1) standard, (2) index patient (hemizygous), (3) sister (heterozygous), (4-13) wildtype male. (C) (1) standard, (2) index patient (hemizygous), (3) sister (heterozygous), (4-12, 14) wildtype female. (D) (1) standard, (2) index patient (hemizygous), (3) sister (heterozygous), (4-15) wildtype male. (E) (1) standard, (2) index patient (hemizygous), (3) sister (heterozygous), (4-6,8,11-14) wildtype female. (F) (1) standard, (2) index patient (hemizygous), (3) sister (heterozygous), (4-14) wildtype male. (G) (1) standard, (2) index patient (hemizygous), (3) sister (heterozygous), (4-7, 12-14) wildtype female, (9) wildtype male. (H) (1) standard, (2) index patient (hemizygous), (3) sister (heterozygous), (4-12) wildtype male, (13-15) wildtype female. (I) (1) standard, (2) index patient (hemizygous), (3) sister (heterozygous), (4, 6-11) wildtype female, (5, 12-15) wildtype male. (J) (1) standard, (2) index patient (hemizygous), (3) sister (heterozygous), (4-11, 13-14) wildtype female. (K) (1) standard, (2) index patient (hemizygous), (3) sister (heterozygous), (4, 6-9,12-15) wildtype male. (L) (1) standard, (2) index patient (hemizygous), (3) sister (heterozygous), (8-11, 14) wildtype female.