**Supplementary Table. Candidate Genes Based on Genetic Inheritance Pattern**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Patient No.** | **Compound Heterozygous** | **De novo or Dominantly Inherited Heterozygous** | **Autosomal recessive** | **X-linked recessive** |
| **Patient 1\*** | 0 | 7^ (*SLC26A1, SEC24A, HRCT1, VIM, ATRNL1, PAOX, NCAPD3*) | 1 (*COL7A1*) | 1 (*ZRSR2*) |
| **Patient 2** | 0 | 0 | 0 | Female |
| **Patient 3** | 6 *( KAT6A, DOCK1, MAN2C1)* | 0 | 1 *(SEMA6C)* | Female |
| **Patient 4** | 0 | 1 (*TCF7L2*) | 0 | Female |
| **Patient 5** | 0 | 1 (***CHD2***) | 0 | 0 |
| **Patient 6\*** |  | 11^ (*IQGAP3, KDM5B, EML6, GULP1, C3orf80, CCRN4L, PSD3, ZNF532, TGM6, MYH9,* ***IGF1R***) | 2 (*ZSCAN12, L3MBTL2*) | 1 (*KIAA1210*) |
| **Patient 7** | 0 | 0 | 0 | Female |
| **Patient 8** | 0 | 0 | 0 | Female |
| **Patient 9\*** | 0 | 7# (*SCL35D1, F13B, PSMD1, PLEC, LURAP1L, RHOT2,* ***IGF1R***) | 0 | Female |

\*Paternal DNA unavailable

^Unable to determine if de novo or paternally inherited

#Novel variant inherited from mother with short stature.

Genes in bold are likely causative genes.