Supplementary Material

**Supplementary Table 1** The potential disease-causing gene variants identified in patients (II-3 and II-5) by whole exome sequencing

| No. | Chromosome/position | Gene symbol | Reference sequence | Variant |
|-----|---------------------|-------------|--------------------|---------|
| 1   | Chr 1/3413552       | MEGF6       | NM_001409.3        | c.3613C>T, p.R1205* |
| 2   | Chr 1/3703514       | LRRC47      | NM_020710.3        | c.976A>G, p.S326G  |
| 3   | Chr 1/23966181      | MDS2        | NM_001348075.2     | c.158T>G, p.L53R   |
| 4   | Chr 1/186359880     | ODR4        | NM_017847.5        | c.512G>T, p.G171V  |
| 5   | Chr 1/206661284     | IKBKE       | XM_005273356.1     | c.1650G>C, p.M550I |
| 6   | Chr 1/247464533     | ZNF496      | XM_005273328.1     | c.1160T>G, p.V387G |
| 7   | Chr 2/17997931      | MSGN1       | NM_001105569.3     | c.146C>T, p.P49L   |
| 8   | Chr 2/112843588     | TMEM87B     | NM_032824.2        | c.845G>A, p.G282D  |
| 9   | Chr 2/179593070     | TTN         | NM_001267550.2     | c.1948T>G, p.L6494R|
| 10  | Chr 2/201369550     | KCTD18      | NM_152387.2        | c.293C>A, p.P98H   |
| 11  | Chr 3/120973797     | STXB5P5L    | NM_014980.2        | c.1497G>T, p.K499N |
| 12  | Chr 3/160156169     | TRIM59      | XM_005247391.1     | c.887C>T, p.P296L  |
| 13  | Chr 4/48552688      | FRYL        | XM_005248082.1     | c.4875C>G, p.N1625K|
| 14  | Chr 4/120213519     | USP53       | NM_019050.2        | c.2375A>G, p.K792R |
| 15  | Chr 4/184627996     | TRAPPC11    | NM_021942.5        | c.3092C>G, p.P1031R|
| 16  | Chr 4/187074881     | FAM149A     | NM_015398.4        | c.169_170insGACCCC |
|     |                     |             |                    | C, p.L60fs        |
| 17  | Chr 5/1037638       | NKD2        | NM_001271082.1     | c.791C>T, p.S264F |
| 18  | Chr 5/52201707      | ITGA1       | NM_181501.1        | c.1424T>A, p.I475N |
| 19  | Chr 5/109178158     | MAN2A1      | NM_002372.4        | c.2966A>G, p.Y899C |
| 20  | Chr 7/1510571       | INTS1       | NM_001080453.2     | c.6368C>G, p.P2123R|
| 21  | Chr 7/97822697      | LMTK2       | NM_014916.4        | c.2920A>G, p.S974G |
| 22  | Chr 7/122338841     | RNF133      | NM_139175.1        | c.132A>G, p.I44M  |
| 23  | Chr 7/150935727     | CHPF2       | XM_005250015.1     | c.2096C>T, p.A699V |
| 24  | Chr 7/151078656     | WDR86       | XM_005249988.1     | c.1207C>T, p.P403S |
| 25  | Chr 8/90926850      | OSGIN2      | NM_001126111.1     | c.404C>G, p.T135R |
| 26  | Chr 9/134397577     | POMT1       | NM_007171.3        | c.2035G>A, p.V679M |
| 27  | Chr 9/138418315     | LCN1        | NM_001252618.1     | c.655delC, p.A212fs|
| 28  | Chr 10/46999656     | GPRIN2      | XM_005270332.1     | c.776C>T, p.P259L  |
| No. | Chromosome/position | Gene symbol* | Reference sequence | Variant |
|-----|---------------------|--------------|-------------------|---------|
| 29  | Chr 10/50013389     | WDFY4        | XM_005270005.1    | c.4569C>G, p.I1523M |
| 30  | Chr 10/102766630    | LZTS2        | NM_032429.4       | c.1715G>C, p.R572P  |
| 31  | Chr 10/135096593    | TUBGCP2      | NM_001256617.1    | c.2362A>G, p.N788D  |
| 32  | Chr 11/22281251     | AN05         | XM_005252822.1    | c.1516T>C, p.F506L  |
| 33  | Chr 11/62554434     | TAF6L        | NM_006473.3       | c.1535C>A, p.S512*  |
| 34  | Chr 12/319017       | SLC6A12      | NM_001122848.2    | c.136G>A, p.V46M    |
| 35  | Chr 12/6127726      | VWF          | NM_000552.5       | c.4858C>T, p.P1620S |
| 36  | Chr 12/48062768     | RPAP3        | NM_024604.3       | c.1644G>C, p.N548K  |
| 37  | Chr 12/50386143     | RACGAP1      | XM_005268811.1    | c.1520A>T, p.H507L  |
| 38  | Chr 12/55795106     | OR6C65       | NM_001005518.1    | c.794G>T, p.G265V   |
| 39  | Chr 12/106532238    | NUAK1        | NM_014840.2       | c.194C>A, p.T65N    |
| 40  | Chr 12/120534220    | RAB35        | XM_005253826.1    | c.816_829del, p.M272fs |
| 41  | Chr 14/45579368     | PRPF39       | NM_017922.3       | c.1248T>A, p.H416Q  |
| 42  | Chr 14/64690070     | SYNE2        | NM_182914.3       | c.2035A>C, p.Q6785P |
| 43  | Chr 14/89016702     | PTPN21       | NM_007039.3       | c.60G>T, p.K20N     |
| 44  | Chr 15/62211567     | VPS13C       | NM_020821.2       | c.7559C>G, p.A2520G |
| 45  | Chr16/1604819       | TMEM204      | NM_001256541.1    | c.473G>A, p.R158K   |
| 46  | Chr 16/2293361      | ECII         | NM_001919.4       | c.521T>C, p.I174T   |
| 47  | Chr 16/67066305     | CES3         | NM_024922.5       | c.1337G>A, p.S446N  |
| 48  | Chr 17/12847454     | ARHGAP44     | NM_014859.4       | c.802A>G, p.I268V   |
| 49  | Chr 17/30648273     | RHBDL3       | XM_005257935.1    | c.1139G>A, p.R380K  |
| 50  | Chr 17/35343961     | AATF         | ENST00000225402.5 | c.878A>G, p.Q293R   |
| 51  | Chr r17/73127637    | NTSC         | NM_001252377.1    | c.166G>C, p.D56H    |
| 52  | Chr 18/2760723      | SMCHD1       | NM_015295.2       | c.4420C>T, p.L1474F |
| 53  | Chr 19/40421117     | FCGBP        | NM_003890.2       | c.2804G>C, p.R935P  |
| 54  | Chr 19/56733965     | ZSCAN5A      | XM_005259254.1    | c.893G>A, p.S298N   |

*Gene symbol is approved by HGNC (HUGO Gene Nomenclature Committee).