***Genetics Research***

**Identification of *FBN1* gene mutations in Ukrainian Marfan syndrome patients**

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**Supplementary Material**

**Supplementary Table S1a. Calcium-binding EGF-like domains of FBN1**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| Start | Sequence | End | Domain | Length |
| 246 | DVDECQ-AIPGLCQG--GNCINTVGSFECKCPAGHKLNEVSQ-KCE | 287 | calcium-binding EGF-like 4 | 42 |
| 288 | DIDECS-TIPGICEG--GECTNTVSSYFCKCPPGFYTSPDGT-RCI | 329 | calcium-binding EGF-like 5 | 42 |
| 490 | DVDECE-KNP--CAG--GECINNQGSYTCQCRAGYQSTLTRT-ECR | 529 | calcium-binding EGF-like 7 | 40 |
| 530 | DIDECL-QNGRICNN--GRCINTDGSFHCVCNAGFHVTRDGK-NCE | 571 | calcium-binding EGF-like 8 | 42 |
| 572 | DMDECSI--RNMCLN--GMCINEDGSFKCICKPGFQLASDGR-YCK | 612 | calcium-binding EGF-like 9 | 41 |
| 613 | DINECET--PGICMN--GRCVNTDGSYRCECFPGLAVGLDGR-VCV | 653 | calcium-binding EGF-like 10 | 41 |
| 723 | DINECA-LDPDICPN--GICENLRGTYKCICNSGYEVDSTGK-NCV | 764 | calcium-binding EGF-like 11 | 42 |
| 765 | DINECV-LNSLLCDN--GQCRNTPGSFVCTCPKGFIYKPDLK-TCE | 806 | calcium-binding EGF-like 12 | 42 |
| 807 | DIDECE-SSP--CIN--GVCKNSPGSFICECSSESTLDPTKT-ICI | 846 | calcium-binding EGF-like 13 | 40 |
| 910 | DIDECE-VFPGVCKN--GLCVNTRGSFKCQCPSGMTLDATGR-ICL | 951 | calcium-binding EGF-like 14 | 42 |
| 1028 | DINECK-MIPSLCTH--GKCRNTIGSFKCRCDSGFALDSEER-NCT | 1069 | calcium-binding EGF-like 15 | 42 |
| 1070 | DIDECR-ISPDLCGR--GQCVNTPGDFECKCDEGYESGFMMMKNCM | 1112 | calcium-binding EGF-like 16 | 43 |
| 1113 | DIDECQ-RDPLLCRG--GVCHNTEGSYRCECPPGHQLSPNIS-ACI | 1154 | calcium-binding EGF-like 17 | 42 |
| 1155 | DINECE-LSAHLCPN--GRCVNLIGKYQCACNPGYHSTPDRL-FCV | 1196 | calcium-binding EGF-like 18 | 42 |
| 1197 | DIDECSI--MNGGCE--TFCTNSEGSYECSCQPGFALMPDQR-SCT | 1237 | calcium-binding EGF-like 19 | 41 |
| 1238 | DIDECE-DNPNICDG--GQCTNIPGEYRCLCYDGFMASEDMK-TCV | 1279 | calcium-binding EGF-like 20 | 42 |
| 1280 | DVNECD-LNPNICLS--GTCENTKGSFICHCDMGYSGKKGKT-GCT | 1321 | calcium-binding EGF-like 21 | 42 |
| 1322 | DINECE-IGAHNCGK-HAVCTNTAGSFKCSCSPGWIGDGIKC-T-- | 1362 | calcium-binding EGF-like 22 | 41 |
| 1363 | DLDECS-NGTHMCSQ-HADCKNTMGSYRCLCKEGYTGDGFTC-T-- | 1403 | calcium-binding EGF-like 23 | 41 |
| 1404 | DLDECS-ENLNLCGN--GQCLNAPGGYRCECDMGFVPSADGK-ACE | 1445 | calcium-binding EGF-like 24 | 42 |
| 1446 | DIDECS-L-PNICVF--GTCHNLPGLFRCECEIGYELDRSGG-NCT | 1486 | calcium-binding EGF-like 25 | 41 |
| 1487 | DVNECLD--PTTCIS--GNCVNTPGSYICDCPPDFELNPTRV-GCV | 1527 | calcium-binding EGF-like 26 | 41 |
| 1606 | DIDECQ-ELPGLCQG--GKCINTFGSFQCRCPTGYYLNEDTR-VCD | 1647 | calcium-binding EGF-like 27 | 42 |
| 1648 | DVNECET--PGICGP--GTCYNTVGNYTCICPPDYMQVNGGN-NCM | 1688 | calcium-binding EGF-like 28 | 41 |
| 1766 | DIDECR-EIPGVCEN--GVCINMVGSFRCECPVGFFYNDKLL-VCE | 1807 | calcium-binding EGF-like 29 | 42 |
| 1808 | DIDECQ-NGPVCQRN--AECINTAGSYRCDCKPGYRFTSTGQ-CN- | 1848 | calcium-binding EGF-like 30 | 41 |
| 1849 | DRNECQ-EIPNICSH--GQCIDTVGSFYCLCHTGFKTNDDQT-MCL | 1890 | calcium-binding EGF-like 31 | 42 |
| 1891 | DINECE-RD--ACGN--GTCRNTIGSFNCRCNHGFILSHNND-CI- | 1929 | calcium-binding EGF-like 32 | 39 |
| 1930 | DVDECASGNGNLCRN--GQCINTVGSFQCQCNEGYEVAPDGR-TCV | 1972 | calcium-binding EGF-like 33 | 43 |
| 1973 | DINECL-LEPRKCAP--GTCQNLDGSYRCICPPGYSLQNEKC-E-- | 2012 | calcium-binding EGF-like 34 | 40 |
| 2013 | DIDECV-EEPEICAL--GTCSNTEGSFKCLCPEGFSLSSSGR-RCQ | 2054 | calcium-binding EGF-like 35 | 42 |
| 2127 | DMDECKE--PDVCKH--GQCINTDGSYRCECPFGYIL--AGN-ECV | 2165 | calcium-binding EGF-like 36 | 39 |
| 2166 | DTDECS-VG-NPCGN--GTCKNVIGGFECTCEEGFEPGPMMT-CE- | 2205 | calcium-binding EGF-like 37 | 40 |
| 2206 | DINECAQ--NPLLCA--FRCVNTYGSYECKCPVGYVLREDRR-MCK | 2246 | calcium-binding EGF-like 38 | 41 |
| 2247 | DEDECE-EGKHDCTEKQMECKNLIGTYMCICGPGYQRRPDGE-GCV | 2290 | calcium-binding EGF-like 39 | 44 |
| 2291 | DENECQ-TKPGICEN--GRCLNTRGSYTCECNDGFTASPNQD-ECL | 2332 | calcium-binding EGF-like 40 | 42 |
| 2402 | DIDECK-VIHDVCRN--GECVNDRGSYHCICKTGYTPDITGT-SCV | 2443 | calcium-binding EGF-like 41 | 42 |
| 2444 | DLNECNQ--APKPCN--FICKNTEGSYQCSCPKGYILQEDGR-SCK | 2484 | calcium-binding EGF-like 42 | 41 |
| 2485 | DLDECA-TKQHNCQF---LCVNTIGGFTCKCPPGFTQHHTSC-I-- | 2523 | calcium-binding EGF-like 43 | 39 |
| 2524 | DNNECTSDINLCGSK--GICQNTPGSFTCECQRGFSLDQTGS-SCE | 2566 | calcium-binding EGF-like 44 | 43 |
| 2567 | DVDECE-GN-HRCQH---GCQNIIGGYRCSCPQGYLQHYQWN-QCV | 2606 | calcium-binding EGF-like 45 | 40 |
| 2607 | DENECLS--AHICGG--ASCHNTLGSYKCMCPAGFQYEQFSG-GCQ | 2647 | calcium-binding EGF-like 46 | 41 |
| 2648 | DINECGS--AQAPCS--YGCSNTEGGYLCGCPPGYFRIGQGH-CV- | 2687 | calcium-binding EGF-like 47 | 40 |
|  | \* :\*\* \* : . : \* \* |  |  |  |

**Supplementary Table S1b. EGF-like domains of FBN1**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| Start | Sequence | End | Domain | Length |
| 81 | ---IVPICRHSCGDGFC-SRPNM--CTCPSGQIAPSCG--- | 112 | EGF-like 1 | 32 |
| 115 | ---SIQHCNIRCMNGGS-CSDDH--CLCQKGYIGTHCG--- | 146 | EGF-like 2 | 32 |
| 147 | ----QPVCESGCLNGGRCVAPNR--CACTYGFTGPQCE--- | 178 | EGF-like 3 | 32 |
| 449 | VTDYCQLVRYLCQNGRCIPTPGSCRCECNKGFQLDLRGECI | 489 | EGF-like 6 | 41 |
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**Supplementary Table S1c. TGFβ-binding domains of FBN1**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| Start | Sequence | End | Domain | Length |
| 184 | GPCFTVI------SNQMCQGQL-SGIVCTKTLCCATV--GRAWGH-PC----EMCPAQPHP--CRRGFI | 236 | TB 1 | 53 |
| 334 | GYCYTAL------TNGRCSNQLPQ--SITKMQCCCDA--GRCWSP-GVTVAPEMCPIRATE--DFNKLC | 389 | TB 2 | 56 |
| 659 | STCYGGY------KRGQCIKPLFG--AVTKSECCCAS--TEYAFGEPC----QPCPAQNSA--EYQALC | 711 | TB 3 | 53 |
| 851 | GTCWQTV------IDGRCEINING--ATLKSQCCSSL--GAAWGS-PC----TLCQVDPICGKGYSR-- | 902 | TB 4 | 52 |
| 956 | ETCFLRY------EDEECTLPIAG--RHRMDACCCSV--GAAWGTEEC----EECPMRNTP--EYEELC | 1008 | TB 5 | 53 |
| 1532 | GNCYLDIRPRGDNGDTACSNEIGV--GVSKASCCCSL--GKAWGT-PC----EMCPAVNTS--EYKILC | 1589 | TB 6 | 58 |
| 1693 | SLCYRNYYAD----NQTCDGELLF--NMTKKMCCCSYNIGRAWNK-PC----EQCPIPSTD--EFATLC | 1748 | TB 7 | 56 |
| 2059 | SYCYAKF------EGGKCSSPKSR--NHSKQECCCAL--KGEGWGDPC----ELCPTEPDE--AFRQIC | 2111 | TB 8 | 53 |
| 2337 | GYCFTEV------LQNMCQIGSSNRNPVTKSECCCDG--GRGWGP-HC----EICPFQGTV--AFKKLC | 2390 | TB 9 | 54 |
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Conserved Cys; X previously described mutation; X novel mutation from this study