***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 |
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**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 |
|  |  \*: \* \*\*. \*  |  |  |  |

 Conserved Cys; X previously described mutation; X novel mutation from this study