Mini-Panels

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| **ردیف** | **نام بیماری** | **تعداد ژن** | **ژن های مورد بررسی** |
| 1 | Afibrinogenemia/ dysfibrinogenemia | 3 | FGA, FGB, FGG |
| 2 | Variable immunodeficiency | 13 | CD19, CD81, CR2, CXCR4, ICOS, LRBA, MS4A1, NFKB1, NFKB2, PRKCD, TNFRSF13B, TNFRSF13C, TNFSF12 |
| 3 | Tuberous sclerosis | 2 | TSC1, TSC2 |
| 4 | Hereditary breast and ovarian cancer (small) | 5 | BRCA1, BRCA2, CHEK2, RAD51C, PALB2 |
| 5 | Hereditary breast and ovarian cancer (large) | 11 | ATM, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53 |
| 6 | Lynch syndrome | 5 | EPCAM, MLH1, MSH2, MSH6, PMS2 |
| 7 | Hereditary hemorrhagic telangiectasia (HHT) | 5 | ACVRL1, ENG, GDF2, RASA1, SMAD4 |
| 8 | Neurofibromatosis | 3 | NF1, NF2, SPRED1 |
| 9 | Hyperekplexia | 3 | GLRA1, GLRB, SLC6A5 |
| 10 | Holoprosencephaly | 9 | CDON, FGFR1, GLI2, PTCH1, SHH, SIX3, TDGF1, TGIF1, ZIC2 |
| 11 | Refsum disease | 8 | AMACR, PEX1, PEX2, PEX26, PEX3, PEX5, PEX7, PHYH |
| 12 | Episodic ataxia | 9 | ATP1A3, CACNA1A, CACNB4, FGF14, KCNA1, KCNQ2, SCN2A, SLC1A3, SLC2A1 |
| 13 | Dopa-responsive dystonia | 3 | GCH1, TH, SPR |
| 14 | Neuropathic pain syndrome | 4 | SCN9A, SCN10A, SCN11A, TRPA1 |
| 15 | Malignant hyperthermia | 3 | RYR1, CACNA1S, STAC3 |
| 16 | Familial intrahepatic cholestasis | 4 | ABCB11, ABCB4, ATP8B1, MYO5B |
| 17 | Maple syrup urine disease | 4 | BCKDHA, BCKDHB, DBT, DLD |
| 18 | MODY syndrome | 12 | ABCC8, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KLF11, NEUROD1, PAX4, PDX1 |
| 19 | Kabuki syndrome | 5 | KDM6A, KMT2D, CHD7, EYA1, IRF6 |
| 20 | Craniosynostosis | 7 | FGFR1, FGFR2, FGFR3, TCF12, TWIST1, ERF, MSX2 |