**پانل Epilepsy، بیماری های متابولیکی و اختلالات تکامل مغزی**

**پانل Epilepsy**

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| **ردیف** | **نام بیماری** | **تعداد ژن** | **ژن های مورد بررسی** |
| 1 | Familial and Idiopathic Epilepsy | 29 | ADRA2B, ALDH7A1, CACNA1A, CHRNA4, CHRNB2, CNTN2, DEPDC5, GABRA1, GABRB3, GABRG2, GRIN2A, KCNA1, KCNMA1, KCNQ2, KCNQ3, KCNT1, LGI1, NPRL2, NPRL3, PRRT2, RELN, SCN1A, SCN1B, SCN2A, SCN8A, SLC1A3, SLC2A1, STX1B, TBC1D24 |
| 2 | Epilepsy and Development Delay | 78 | AARS, ALDH7A1, ALG13, AMT, ARHGEF9, ARX, BRAT1, CACNA1A, CASK, CDKL5, CHD2, CNNM2, DNM1, DOCK7, EEF1A2, FOXG1, GABRA1, GABRB3, GAMT, GLDC, GNAO1, GNB1, GPHN, GRIN1, GRIN2A, GRIN2B, GRIN2D, HACE1, HCN1, HNRNPU, IQSEC2, ITPA, KCNA2, KCNB1, KCNQ2, KCNT1, MBD5, MECP2, MEF2C, MOCS1, MOCS2, NECAP1, PCDH19, PIGA, PLCB1, PNKP, PNPO, POLG, PURA, QARS, ROGDI, SCN1A, SCN2A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A8, SLC9A6, SMC1A, SPTAN1, ST3GAL3, ST3GAL5, STXBP1, SYNGAP1, SZT2, TBC1D24, TCF4, TSC1, TSC2, UBE3A, WDR45, WWOX, ZEB2 |
| 3 | Neuronal Ceroid Lipofuscinosis and Progressive Myoclonus Epilepsy | 28 | AFG3L2, ASAH1, ATP13A2, CARS2, CERS1, CLN3, CLN5, CLN6, CLN8, CSTB, CTSD, CTSF, DNAJC5, EPM2A, GOSR2, GRN, KCNC1, KCTD7, LMNB2, MFSD8, NEU1, NHLRC1, PPT1, PRDM8, PRICKLE1, SCARB2, SERPINI1, TPP1 |
| 4 | GPI anchor deficiency with or without Hyperphosphatasia | 12 | PGAP2, PGAP3, PIGA, PIGG, PIGL, PIGM, PIGN, PIGO, PIGT, PIGV, PIGW, PIGY |
| 5 | Migraine | 9 | ATP1A2, ATP1A3, CACNA1A, NOTCH3, POLG, PRRT2, SCN1A, SLC1A3, SLC2A1 |
| 6 | Hyperekplexia | 3 | GLRA1, GLRB, SLC6A5 |
| 7 | Metabolic/ Mitochondrial Epilepsy | 92 | AARS2, ABAT, ABCC8, ACY1, ADK, ADSL, ALDH5A1, ALDH7A1, AMT, ATIC, AUH, BCKDHA, BCKDHB, BCKDK, BCS1L, BTD, CARS2, CNNM2, COQ4, COQ8A, CPT2, D2HGDH, DARS2, DBT, DHFR, DLD, DPYD, EARS2, ETFA, ETFB, ETFDH, ETHE1, FARS2, FH, FOLR1, FOXRED1, GAMT, GATM, GCDH, GCH1, GCK, GCSH, GFM1, GLDC, GLUD1, GLUL, GPHN, HADH, HLCS, HPD, IDH2, ITPA, IVD, KCNJ11, L2HGDH, LIAS, MLYCD, MMACHC, MOCS1, MOCS2, MT-ATP6 (m.8993T>G/C), MT-TK (m.8344A>G), MT-TL1 (m.3243A>G; m.3271T>C), MTHFR, NARS2, NDUFA1, PC, PCBD1, PCCA, PCCB, PDHA1, PDHX, PDSS2, PET100, PHGDH, PNPO, POLG, PSAT1, PSPH, PTS, QDPR, SDHA, SLC16A1, SLC19A3, SLC25A1, SLC2A1, SLC46A1, SLC6A8, SUOX, SURF1, TWNK, VARS2 |

**پانل بیماری های متابولیکی**

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| **ردیف** | **نام بیماری** | **تعداد ژن** | **ژن های مورد بررسی** |
| 1 | Congenital Disorders of Glycosylation (CDG syndrome) | 44 | ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, B4GALT1, CAD, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DDOST, DOLK, DPAGT1, DPM1, DPM2, DPM3, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NGLY1, PGM1, PMM2, RFT1, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, STT3A, STT3B, TMEM165, TMEM199 |
| 2 | Lysosomal Disorders | 37 | AGA, ARSA, ARSB, CTNS, CTSA, FUCA1, GALC, GALNS, GBA, GLA, GLB1, GM2A, GNPTAB, GNPTG, GNS, GUSB, HEXA, HEXB, HGSNAT, HYAL1, IDS, IDUA, LIPA, MAN1B1, MAN2B1, MANBA, MCOLN1, NAGA, NAGLU, NEU1, NPC1, NPC2, PSAP, SGSH, SLC17A5, SMPD1, SUMF1 |
| 3 | Peroxisome Biogenesis Disorders: Zellweger Syndrome Spectrum | 19 | ABCD1, ACOX1, AMACR, HSD17B4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, SCP2 |
| 4 | Pyridoxine and Folic Acid dependent Epilepsy | 6 | ALDH7A1, DHFR, FOLR1, MTHFR, PNPO, SLC46A1 |
| 5 | Glutaraciduria | 4 | ETFA, ETFB, ETFDH, GCDH |
| 6 | Glycine Encephalopathy | 3 | AMT, GCSH, GLDC |
| 7 | Hyperphenylalaninemia | 5 | GCH1, PAH, PCBD1, PTS, QDPR |
| 8 | Maple Syrup Urine Disease and DLD Deficiency | 4 | BCKDHA, BCKDHB, DBT, DLD |
| 9 | Molybdenum Cofactor Deficiency and Sulfite Oxidase Deficiency | 4 | GPHN, MOCS1, MOCS2, SUOX |
| 10 | Urea Cycle Disorders and Secondary Hyperammonemia | 18 | ARG1, ASL, ASS1, CPS1, CPT2, GLUD1, LMBRD1, MMAA, MMAB, MMACHC, MMADHC, MUT, NAGS, OTC, PCCA, PCCB, SLC25A15, SLC7A7 |
| 11 | Glycogen Storage Disease | 20 | AGL, ALDOA, FBP1, G6PC, GAA, GBE1, GYS1, GYS2, LAMP2, LDHA, PFKM, PGAM2, PHKA1, PHKA2, PHKB, PHKG2, PYGL, PYGM, SLC2A2, SLC37A4 |
| 12 | Hyperinsulinemic Hypoglycemia | 7 | ABCC8, GCK, GLUD1, HADH, INSR, KCNJ11, SLC16A1 |
| 13 | Methylmalonic Acidemia | 6 | LMBRD1, MMAA, MMAB, MMACHC, MMADHC, MUT |

**پانل اختلالات تکامل مغزی**

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| **ردیف** | **نام بیماری** | **تعداد ژن** | **ژن های مورد بررسی** |
| 1 | Microcephaly and Pontocerebellar Hypoplasia | 64 | AMPD2, ANKLE2, ASNS, ASPM, ATR, BRF1, CASK, CDC6, CDK5RAP2, CDK6, CDT1, CENPE, CENPJ, CEP135, CEP152, CEP63, CHMP1A, CKAP2L, CLP1, DYRK1A, EFTUD2, EXOSC3, EXOSC8, FOXG1, GMNN, IER3IP1, KAT6A, KIF11, KNL1, MBD5, MCPH1, MFSD2A, NIN, ORC1, ORC4, ORC6, PCLO, PHC1, PLK4, PNKP, PPP1R15B, PQBP1, QARS, RARS2, RBBP8, SASS6, SEPSECS, SLC25A19, SMARCA2, SPATA5, STAMBP, STIL, TRAIP, TRMT10A, TSEN2, TSEN34, TSEN54, TUBGCP4, TUBGCP6, VPS53, VRK1, WDR62, WDR73, ZNF335 |
| 2 | Neuronal Migration Disorders | 66 | ACTB, ACTG1, ADGRG1, AKT3, ARFGEF2, ARX, B3GALNT2, B3GNT1, CCND2, CDK5, COL4A1, COL4A2, DAG1, DCHS1, DCX, DYNC1H1, EMX2, EOMES, ERMARD, FAT4, FKRP, FKTN, FLNA, GMPPB, IER3IP1, ISPD, KATNB1, KIF1BP, KIF2A, KIF5C, LAMB1, LAMC3, LARGE, MEF2C, MTOR, NDE1, NEDD4L, OCLN, PAFAH1B1, PI4KA, PIK3CA, PIK3R2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RAB18, RAB3GAP1, RAB3GAP2, RELN, RTTN, SHH, SIX3, TBC1D20, TMEM5, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, VLDLR, WDR62, WDR81 |
| 3 | Holoprosencephaly Spectrum | 9 | CDON, FGFR1, GLI2, PTCH1, SHH, SIX3, TDGF1, TGIF1, ZIC2 |
| 4 | Macrocephaly | 33 | AKT3, ASPA, BRWD3, CCDC88C, CCND2, CUL4B, DNMT3A, EZH2, GCDH, GFAP, GPC3, HEPACAM, IGF2, KPTN, L1CAM, MED12, MLC1, MTOR, NFIX, NSD1, PHF6, PIGA, PIGN, PIGT, PIK3CA, PIK3R2, PTCH1, PTEN, RIN2, RNF135, SETD2, STRADA, TBC1D7 |
| 5 | Leukodystrophy and Leukoencephalopathy | 76 | AARS, AARS2, ABCD1, ACOX1, ADAR, AIMP1, ALDH3A2, ARSA, ASPA, BCAP31, C11orf73, CLCN2, CSF1R, CTC1, CYP27A1, DARS, DARS2, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, FAM126A, FUCA1, GALC, GBE1, GCDH, GFAP, GJC2, HEPACAM, HSD17B4, HSPD1, HTRA1, IFIH1, L2HGDH, LMNB1, MLC1, NOTCH3, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PLEKHG2, PLP1, POLR1C, POLR3A, POLR3B, PSAP, PYCR2, RARS, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SCP2, SLC16A2, SLC17A5, SOX10, SUMF1, TREM2, TREX1, TUBB4A, TYROBP, VPS11 |
| 6 | Aicrdi-Goutieres Syndrome | 7 | ADAR, IFIH1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1 |
| 7 | Joubert Syndrome/ Meckel Syndrome | 30 | AHI1, ARL13B, B9D1, B9D2, C5orf42, CC2D2A, CEP104, CEP164, CEP290, CEP41, CSPP1, INPP5E, KIAA0586, KIF7, MKS1, NPHP1, NPHP3, OFD1, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423 |
| 8 | Cornelia de lange Syndrome | 6 | HDAC8, NIPBL, RAD21, SMC1A, SMC3, UBE2A |
| 9 | Cerebral Microangiopathies | 6 | COL4A1, CTC1, GLA, HTRA1, NOTCH3, TREX1 |
| 10 | Leukodystrophy and Leukoencephalopathy and Differential Diagnosis | 176 | AARS, AARS2, ABCD1, ACOX1, ADAR, AGA, AIMP1, ALDH3A2, AP4E1, APOPT1, ARSA, ASPA, ATP7A, ATP7B, AUH, BCAP31, BCS1L, C11orf73, C12orf65, CLCN2, COL4A1, COL4A2, COX10, COX15, COX6B1, CSF1R, CTC1, CYP27A1, CYP7B1, D2HGDH, DARS, DARS2, DDHD2, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ERCC2, ERCC3, ERCC6, ERCC8, ETFA, ETFB, ETFDH, FA2H, FAM126A, FARS2, FKRP, FKTN, FOLR1, FOXRED1, FUCA1, GALC, GAN, GBE1, GCDH, GFAP, GFM1, GJC2, GLA, GLB1, GMPPB, HEPACAM, HEXA, HMGCL, HSD17B4, HSPD1, HTRA1, IBA57, IDS, IFIH1, ISCA2, KCNT1, L2HGDH, LAMA2, LARGE, LMNB1, LRPPRC, MCOLN1, MLC1, MMACHC, MPV17, MTFMT, NADK2, NDUFA1, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF5, NDUFAF6, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NEU1, NOTCH3, NPC1, NPC2, NUBPL, OCLN, OCRL, PC, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHGDH, PHYH, PLA2G6, PLEKHG2, PLP1, POLG, POLR1C, POLR3A, POLR3B, POMGNT1, POMT1, POMT2, PRF1, PSAP, PSAT1, PYCR2, RARS, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SCO2, SCP2, SDHA, SDHAF1, SLC16A2, SLC17A5, SLC25A1, SLC25A12, SOX10, SPG11, SPG20, SPG21, STX11, STXBP2, SUCLA2, SUMF1, SURF1, TACO1, TBCK, TMEM70, TREM2, TREX1, TTC19, TUBB4A, TUFM, TYMP, TYROBP, UNC13D, VPS11, ZFYVE26 |

**پانل Epilepsy، بیماری های متابولیکی و اختلالات تکامل مغزی**

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| **ردیف** | **نام بیماری** | **تعداد ژن** | **ژن های مورد بررسی** |
| 1 | Complete Panel- Epilepsy, Metabolic and Brain Development Disorders | 634 | AARS, AARS2, ABAT, ABCC8, ABCD1, ACOX1, ACTB, ACTG1, ACY1, ADAR, ADGRG1, ADK, ADRA2B, ADSL, AFG3L2, AGA, AGL, AHI1, AIMP1, AKT3, ALDH3A2, ALDH5A1, ALDH7A1, ALDOA, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, AMACR, AMPD2, AMT, ANKLE2, AP4E1, APOPT1, ARFGEF2, ARG1, ARHGEF9, ARL13B, ARSA, ARSB, ARX, ASAH1, ASL, ASNS, ASPA, ASPM, ASS1, ATIC, ATP13A2, ATP1A2, ATP1A3, ATP7A, ATP7B, ATR, AUH, B3GALNT2, B3GNT1, B4GALT1, B9D1, B9D2, BCAP31, BCKDHA, BCKDHB, BCKDK, BCS1L, BRAT1, BRF1, BRWD3, BTD, C12orf65, C5orf42, CACNA1A, CAD, CARS2, CASK, CC2D2A, CCDC115, CCDC88C, CCND2, CDC6, CDK5, CDK5RAP2, CDK6, CDKL5, CDON, CDT1, CENPE, CENPJ, CEP104, CEP135, CEP152, CEP164, CEP290, CEP41, CEP63, CERS1, CHD2, CHMP1A, CHRNA4, CHRNB2, CKAP2L, CLCN2, CLN3, CLN5, CLN6, CLN8, CLP1, CNNM2, CNTN2, COG1, COG2, COG4, COG5, COG6, COG7, COG8, COL4A1, COL4A2, COQ4, COQ8A, COX10, COX15, COX6B1, CPS1, CPT2, CSF1R, CSPP1, CSTB, CTC1, CTNS, CTSA, CTSD, CTSF, CUL4B, CYP27A1, CYP7B1, D2HGDH, DAG1, DARS, DARS2, DBT, DCHS1, DCX, DDHD2, DDOST, DEPDC5, DHFR, DLD, DNAJC5, DNM1, DNMT3A, DOCK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DYNC1H1, DYRK1A, EARS2, EEF1A2, EFTUD2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EMX2, EOMES, EPM2A, ERCC2, ERCC3, ERCC6, ERCC8, ERMARD, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, EXOSC8, EZH2, FA2H, FAM126A, FARS2, FAT4, FBP1, FGFR1, FH, FKRP, FKTN, FLNA, FOLR1, FOXG1, FOXRED1, FUCA1, G6PC, GAA, GABRA1, GABRB3, GABRG2, GALC, GALNS, GAMT, GAN, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GFAP, GFM1, GJC2, GLA, GLB1, GLDC, GLI2, GLRA1, GLRB, GLUD1, GLUL, GM2A, GMNN, GMPPB, GNAO1, GNB1, GNPTAB, GNPTG, GNS, GOSR2, GPC3, GPHN, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRN, GUSB, GYS1, GYS2, HACE1, HADH, HCN1, HDAC8, HEPACAM, HEXA, HEXB, HGSNAT, HIKESHI, HLCS, HMGCL, HNRNPU, HPD, HSD17B4, HSPD1, HTRA1, HYAL1, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, IGF2, INPP5E, INSR, IQSEC2, ISCA2, ISPD, ITPA, IVD, KAT6A, KATNB1, KCNA1, KCNA2, KCNB1, KCNC1, KCNJ11, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7, KIAA0586, KIF11, KIF1BP, KIF2A, KIF5C, KIF7, KNL1, KPTN, L1CAM, L2HGDH, LAMA2, LAMB1, LAMC3, LAMP2, LARGE, LDHA, LGI1, LIAS, LIPA, LMBRD1, LMNB1, LMNB2, LRPPRC, MAN1B1, MAN2B1, MANBA, MBD5, MCOLN1, MCPH1, MECP2, MED12, MEF2C, MFSD2A, MFSD8, MGAT2, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MOCS2, MOGS, MPDU1, MPI, MPV17, MT-ATP6 (m.8993T>G/C), MT-TK (m.8344A>G), MT-TL1 (m.3243A>G; m.3271T>C), MTFMT, MTHFR, MTOR, MUT, NADK2, NAGA, NAGLU, NAGS, NARS2, NDE1, NDUFA1, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF5, NDUFAF6, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NECAP1, NEDD4L, NEU1, NFIX, NGLY1, NHLRC1, NIN, NIPBL, NOTCH3, NPC1, NPC2, NPHP1, NPHP3, NPRL2, NPRL3, NSD1, NUBPL, OCLN, OCRL, OFD1, ORC1, ORC4, ORC6, OTC, PAFAH1B1, PAH, PC, PCBD1, PCCA, PCCB, PCDH19, PCLO, PDE6D, PDHA1, PDHX, PDSS2, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGAP2, PGAP3, PGM1, PHC1, PHF6, PHGDH, PHKA1, PHKA2, PHKB, PHKG2, PHYH, PI4KA, PIGA, PIGG, PIGL, PIGM, PIGN, PIGO, PIGT, PIGV, PIGW, PIGY, PIK3CA, PIK3R2, PLA2G6, PLCB1, PLEKHG2, PLK4, PLP1, PMM2, PNKP, PNPO, POLG, POLR1C, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPP1R15B, PPT1, PQBP1, PRDM8, PRF1, PRICKLE1, PRRT2, PSAP, PSAT1, PSPH, PTCH1, PTEN, PTS, PURA, PYCR2, PYGL, PYGM, QARS, QDPR, RAB18, RAB3GAP1, RAB3GAP2, RAD21, RARS, RARS2, RBBP8, RELN, RFT1, RIN2, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF135, ROGDI, RPGRIP1L, RTTN, SAMHD1, SASS6, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SCO2, SCP2, SDHA, SDHAF1, SEPSECS, SERPINI1, SETD2, SGSH, SHH, SIK1, SIX3, SLC12A5, SLC13A5, SLC16A1, SLC16A2, SLC17A5, SLC19A3, SLC1A3, SLC25A1, SLC25A12, SLC25A15, SLC25A19, SLC25A22, SLC2A1, SLC2A2, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A8, SLC46A1, SLC6A1, SLC6A5, SLC6A8, SLC7A7, SLC9A6, SMARCA2, SMC1A, SMC3, SMPD1, SOX10, SPATA5, SPG11, SPG20, SPG21, SPTAN1, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STAMBP, STIL, STRADA, STT3A, STT3B, STX11, STX1B, STXBP1, STXBP2, SUCLA2, SUMF1, SUOX, SURF1, SYNGAP1, SZT2, TACO1, TBC1D20, TBC1D24, TBC1D7, TBCK, TCF4, TCTN1, TCTN2, TCTN3, TDGF1, TGIF1, TMEM138, TMEM165, TMEM199, TMEM216, TMEM231, TMEM237, TMEM5, TMEM67, TMEM70, TPP1, TRAIP, TREM2, TREX1, TRMT10A, TSC1, TSC2, TSEN2, TSEN34, TSEN54, TTC19, TTC21B, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP4, TUBGCP6, TUFM, TWNK, TYMP, TYROBP, UBE2A, UBE3A, UNC13D, VARS2, VLDLR, VPS11, VPS53, VRK1, WDR45, WDR62, WDR73, WDR81, WWOX, ZEB2, ZFYVE26, ZIC2, ZNF335, ZNF423 |