

Order Form

CeGaT GmbH
Praxis für Humangenetik
Paul-Ehrlich Str. 17
72076 Tübingen

CeGaT-Panels

Epilepsy, Migraine, Metabolic disorders and
Syndromal disorders with epilepsy



Patient / Person

Surname:

First name:

Date of birth:

DNA-No.:

Sent by

Name:

Mailing address:

Phone number:

Date & Signature:

Indication / Diagnosis

<input type="checkbox"/> autosomal dominant	<input type="checkbox"/> sporadic
<input type="checkbox"/> autosomal recessive	<input type="checkbox"/> familial
<input type="checkbox"/> X-chromosomal	<input type="checkbox"/> consanguin
<input type="checkbox"/> unclear	<input type="checkbox"/> segregation

Material

Invoice

to patient

to sender

Panels

Panel 1: Generalized / Myoclonic Epilepsy, Febrile Seizures; Absences: ALDH7A1, BRD2, CACNA1A, CACNA1H, CACNB4, CASR, CHRNA2, CHRNA4, CHRN2, CLCN2, CSTB, EFHC1, EPM2A, GABRA1, GABRB3, GABRD, GABRG2, GPR98, RIN2A, GRIN2B, KCNMA1, KCNQ2, KCNQ3, KCTD7, MBD5, ME2, NHLRC1, PCDH19, PRICKLE1, PRICKL2, SCARB2, SCN1A, SCN1B, SCN2A, SCN9A, SLC2A1, TBCQD24

Panel 2: Epileptic Encephalopathies: ARGHGEF9, ARX, CDKL5, CNTNAP2, FOXG1, GABRG2, GRIN2A, GRIN2B, MAPK10, MECP2, NRXN1, PCDH19, PNKP, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SCN1A, SCN1B, SCN2A, SCN9A, SLC2A1, SLC25A22, SLC9A6, SPTAN1, STXBP1, TCF4, TREX1, UBE3A, ZEB2

Panel 3: Epilepsy and X-linked Mental Retardation: ARGHGEF9, ARX, ATP6AP2, ETRX, CASK, CDKL5, CUL4B, CXORF5, DCX, FGD1, GPC3, GRIA3, HSD17B10, JARID1C, OPHN1, PAK3, PHF6, PLP1, PQBP1, RAB39B, SLC9A6, SMC1A, SMS, SRPX2, SYP

Panel 4: CDG Syndrome: ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG12, B4GALT1, COG1, COG7, COG8, DPAGT1, DPM1, DPM3, GCS1, MGAT2, MPDU1, MPI, PMM2, RFT1, SLC35A1, SLC35C1, TMEM15

Panel 5: Ceroidlipofuscinosis: CLN3, CLN5, CLN6, CLN8, CTSD, MFSD8, PPT1, TPP1

Panel 6: Coenzyme Q Deficiency Syndrome: APTX, CABC1 (ADCK3), COQ9, PDSS1, PDSS2

Panel 7: Joubert-Syndrome: AHI1, ARL13B, CC2D2A, CEP290, CXORF5, INPP5E, NPHP1, RPRG1P1L, TMEM67, TMEM126

Panel 8: Selected Mitochondrial Disorders: APTX, ATPAF2, BCS1L, C12ORF65, C8ORF38, CABC1 (ADCK3), COQ2, COQ9, COX10, COX15, DLD, GCSH, GCST, GLDC, HSD17B10, LRPPRC, NDUFA2, NDUFS1, NDUFS3, NDUFS4, NDUFS7, NDUFS8, NDUFV1, PC, PDHA1, PSDD1, PDSS2, POLG, RARS2, SCD2, SDHA, SURF1, TACO1, TMEM70, VDAC1

Panel 9: Lissencephaly and Polymicrogyria: COL18A1, CPT2, DCX, EOMES, FGFR3, FLNA, GPR56, PAFAH1B1, PAX6, PEX7, RAB3GAP1, RELN, SNAP29, SRPX2, TUBA1A, TUBA8, TUBB2B, VDAC1

Panel 10: Severe Microcephaly and Pontocerebellar Hypoplasia: ASPM, ATR, BUB1B, CASK, CDK5RAP2, CENPJ, CEP152, LIG4, MCPH1, MED17, NHEJ1, PCNT, PNKP, PQBP1, RARS2, SLC25A19, STIL, TSEN2, TSEN34, TSEN54, VRK1, WDR62

Panel 11: MPS and Mucopolidosis: ARSB, GALNS, GLB1, GNPTAB, GNPTG, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, MCOLN1, NAGLU, SGSH, SUMF1

Panel 12: Disorders of the Ras-MAPK Pathway: BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, MYSD4, NF1, NRAS, PTPN11, RAF1, SHOC2, SOS1, SPRED1

Panel 13: Walker-Warburg Syndrome: FKRP, FKTN, LARGE, POMGNT1, POMT1, POMT2

Panel 14: Disorders of Peroxisome Biogenesis: PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX12, PEX14, PEX26

Panel 15: Metabolic Disorders: ABCC8, ACY1, ADSL, AGA, ALDH4A1, ALDH5A1, ALDH7A1, ARG1, ARSA, ASPA, ATIC, BTBD, CPT2, CTSA, DPYD, ETFB, ETLFDH, FH, FOLR1, FUCA1, GALC, GAMT, GCDH, GCSH, GCST, GLB1, GLDC, GNE, HEXA, HEXB, HPD, L2HGDH, LAMA2, MOCS1, MOCS2, NEU1, NPC1, NPC2, PGK1, PRODH, PSAP, QDPR, SLC17A5, SLC25A15, SLC46A1, SMPD1, SUMF1, SUOX

Panel 16: Leukodystrophies: ARSA, ASPA, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, GALC, GFAP, MLC1, NOTCH3, PLP1, PSAP, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SDHA, SUMF1, TREX1

Panel 17: Migraine: ATP1A2, CACNA1A, NOTCH3, POLG, SCN1A, SLC2A1

Panel 18: Hyperekplexia: ARGHGEF9, GLRA1, GLRB, GPHN, SLC6A5

Panel 19: Holoprosencephaly: FGF8, GLI2, GLI3, PTCH1, SHH, SIX3, TGIF, ZIC2

Panel 20: Neuronal Migration Disorders: ARFGEF2, ARX, COL18A1, COL4A1, CPT2, DCX, EMX2, EOMES, FGFR3, FKRP, FKTN, FLNA, GRP56, LAMA2, LARGE, PAFAH1B1, PAX6, PEX7, POMGNT1, POMT1, POMT2, PQBP1, RAB3GAPRELN, SNAP29, SRPX2, TUBA1A, TUBA8, TUBB2B, VDAC1, WDR62

Other Syndromal Disorders with Epilepsy: ARFGEF2, ARGHGEF9, A2BP1, ASPA, ATP1A2, ATP2A2, ATP6V0A2, CACNA1A, CCDC88C, CLCNKA, CLCNKB, COH1, DLGAP2, GFAP, GLI3, GLRA1, GLRB, GPHN, KCNA1, KCNJ1, KCNJ10, KIAA1279, LAMA2, LBR, LGI1, MLC1, MLL2, NF1, NIPBL, PANK2, PI12, PIGV, PLA2G6, RAI1, SCN8A, SETBP1, SHH, SLC4A10, SLC6A5, SMC1A, SMC3, SYNGAP1, TBX1, TSC1, TSC2, VPS13A, ZEB2

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Declaration of consent

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Patient / Person Medical doctor

Anonymization for scientific purposes

I agree that left over material might be anonymously used for research purposes

yes no Patient / Person