NIPD GENETICS

Ventrilia

cardiovascular test

CARDIOVASCULAR PANELS

WHAT IS THE ROLE OF CARDIOVASCULAR GENETIC TESTING?

IDENTIFY

- Genetic mutations involved in cardiovascular conditions
- Complex cardiovascular diseases
- The correct disease by differentiating between diseases with similar phenotype

EVALUATE

- Patient's level of risk
- Treatment options
- At-risk family members

MANAGE

- Clinical care
- Decision-making before an invasive treatment
- Specific therapies and predict their response

7 INDIVIDUAL PANELS AND 1 COMPREHENSIVE PANEL FOR INHERITED CARDIOVASCULAR DISEASES

AORTOPATHY (48 genes)							
ABCC6	*ATP6V1A*	*C1S*	*COL4A1*	*FBLN5*	*LOX*	*PRDM5*	*SMAD4*
ACTA2	*ATP6V1E1*	*CBS*	*COL5A1*	*FBN1*	*LTBP4*	*PYCR1*	*TGFB2*
ACVR1	*B3GALT6*	*CHST14*	*COL5A2*	*FBN2*	*MFAP5*	*SKI*	*TGFB3*
ADAMTS2	*B4GALT7*	*COL1A1*	*DSE*	*FKBP14*	*MYH11*	*SLC2A10*	*TGFBR1*
ALDH18A1	*BGN*	*COL1A2*	*EFEMP2*	*FOXE3*	*MYLK*	*SLC39A13*	*TGFBR2*
ATP6V0A2	*C1R*	*COL3A1*	*ELN*	*GORAB*	*PLOD1*	*SMAD3*	*ZNF469*

ARRHYTHMIA (42 genes)						
ABCC9	*CALM1*	*GNAI2*	*KCNE1*	*KCNJ8*	*SCN10A*	*SNTA1*
AKAP9	*CALM2*	*GPD1L*	*KCNE2*	*KCNQ1*	*SCN1B*	*TBX5*
ANK2	*CALM3*	*HCN4*	*KCNE3*	*LMNA*	*SCN2B*	*TECRL*
CACNA1C	*CASQ2*	*KCNA5*	*KCNH2*	*MYL4*	*SCN3B*	*TNNI3*
CACNA2D1	*CAV3*	*KCNAB2*	*KCNJ2*	*NPPA*	*SCN4B*	*TRDN*
CACNB2	*GJA5*	*KCND3*	*KCNJ5*	*RYR2*	*SCN5A*	*TRPM4*

CARDIOMYOPATHY (98 genes)

AARS2	*CAV3*	*DSP*	*HCN4*	*MYBPC3*	*PLN*	*TAZ*
ABCC9	*COA5*	*DTNA*	*HRAS*	*MYH6*	*PPA2*	*TCAP*
ACAD9	*COA6*	*ELAC2*	*ILK*	*MYH7*	*PRDM16*	*TGFB3*
ACADVL	*COX15*	*EMD*	*JPH2*	*MYL2*	*PRKAG2*	*TK2*
ACTA1	*CPT2*	*FHL1*	*JUP*	*MYL3*	*RAF1*	*TMEM43*
ACTC1	*CRYAB*	*FKTN*	*KARS*	*MYLK2*	*RBM20*	*TMEM70*
ACTN2	*CSRP3*	*FLNC*	*LAMA4*	*MYOZ2*	*RYR2*	*TNNC1*
AGK	*CTNNA3*	*GAA*	*LAMP2*	*MYPN*	*SCN5A*	*TNNI3*
AKAP9	*DES*	*GATAD1*	*LDB3*	*NDUFB11*	*SCO2*	*TNNT2*
ALMS1	*DMD*	*GBE1*	*LMNA*	*NDUFV2*	*SGCD*	*TPM1*
ALPK3	*DNAJC19*	*GLA*	*MIB1*	*NEBL*	*SLC22A5*	*TSFM*
ANKRD1	*DOLK*	*GTPBP3*	*MRPL3*	*NEXN*	*SLC25A20*	*TTN*
BAG3	*DSC2*	*HADHA*	*MRPL44*	*PDLIM3*	*SLC25A3*	*TTR*
CASQ2	*DSG2*	*HADHB*	*MTO1*	*PKP2*	*SLC25A4*	*VCL*

CONGENITAL HEART DEFECTS (80 genes)

ACTA2	*CELSR2*	*DNAH6*	*FOXC1*	*HAND2*	*MYH7*	*PITX2*	*SALL4*	*TGFBR1*
ACTC1	*CELSR3*	*DNAI1*	*FOXH1*	*HRAS*	*NIPBL*	*PKD1L1*	*SEMA3D*	*TGFBR2*
ACVR2B	*CHD4*	*DTNA*	*GANAB*	*JAG1*	*NKX2-5*	*PLD1*	*SEMA3E*	*TLL1*
AFF4	*CHD7*	*EHMT1*	*GATA4*	*KDM5B*	*NKX2-6*	*POGZ*	*SMAD6*	*TMEM260*
BMPR2	*CITED2*	*ELN*	*GATA5*	*KMT2D*	*NME7*	*PRDM6*	*TAB2*	*TPM1*
CAD	*CREBBP*	*EVC*	*GATA6*	*MED13L*	*NODAL*	*PRKD1*	*TBX1*	*ZEB2*
CCDC11	*CRELD1*	*EVC2*	*GDF1*	*MMP21*	*NOTCH1*	*RABGAP1L*	*TBX20*	*ZFPM2*
CDK13	*DNAH11*	*FBN1*	*GPC3*	*MYH11*	*NOTCH2*	*RBFOX2*	*TBX5*	*ZIC3*
CELSR1	*DNAH5*	*FLNA*	*HAND1*	*MYH6*	*NR2F2*	*RBM10*	*TFAP2B*	

FAMILIAL HYPERCHOLESTEROLEMIA (11 genes)

ABCA1	*APOA5*	*LDLR*	*LPL*
ABCG5	*APOB*	*LDLRAP1*	*PCSK9*
ABCG8	*APOE*	*LIPA*	

PULMONARY HYPERTENSION (11 genes)

ACVRL1	*CAV1*	*KCNA5*	*SMAD9*
BMPR1B	*EIF2AK4*	*KCNK3*	*TBX4*
BMPR2	*ENG*	*SMAD4*	

RASOPATHIES (30 genes)

AKT3	*CCND2*	*KRAS*	*MAP2K2*	*NF2*	*PIK3R2*	*RAF1*	*RIT1*	*SHOC2*	*SOS2*
BRAF	*EPHB4*	*LZTR1*	*MRAS*	*NRAS*	*PPP1CB*	*RASA1*	*RRAS*	*SMARCB1*	*SPRED1*
CBL	*HRAS*	*MAP2K1*	*NF1*	*PIK3CA*	*PTPN11*	*RASA2*	*SASH1*	*SOS1*	*STAMBP*

COMPREHENSIVE PANEL (292 genes)

Includes all 292 genes

NIPD GENETICS

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