Roadmap 2 Rare

Full table of genes and disorders - 60-gene HCM panel

GENE	DISORDER
A2ML1	Noonan syndrome
ACADVL	VLCAD deficiency
ACTC1	Atrial septal defect 5; cardiomyopathy, dilated, 1R; cardiomyopathy, hypertrophic, 11; left ventricular noncompaction 4
ACTN2	Cardiomyopathy, dilated, 1AA, with or without LVNC; cardiomyopathy, hypertrophic, 23, with or without LVNC; myopathy, congenital with structured cores and Z-line abnormalities; myopathy, distal, 6, adult onset
AGL	Glycogen storage disease IIIa; glycogen storage disease IIIb
ANKRD1	KBG syndrome, cardiomyopathy, dilated; cardiomyopathy, hypertrophic
BAG3	Cardiomyopathy, dilated, 1HH; myopathy, myofibrillar, 6
BRAF	Adenocarcinoma of lung, somatic; cardiofaciocutaneous syndrome; colorectal cancer, somatic; LEOPARD syndrome 3; melanoma, malignant, somatic; non-small cell lung cancer, somatic; Noonan syndrome 7
CACNA1C	Brugada syndrome 3; long QT syndrome 8; Timothy syndrome
CALR3	Cardiomyopathy, hypertrophic
CAV3	Cardiomyopathy, familial hypertrophic; creatine phosphokinase, elevated serum; long QT syndrome 9; myopathy, distal, Tateyama type; rippling muscle disease 2
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia
CPT2	CPT II deficiency, infantile; CPT II deficiency, lethal neonatal; CPT II deficiency, myopathic, stress-induced
CSRP3	Cardiomyopathy, hypertrophic, 12
DES	Cardiomyopathy, dilated, 1I; myopathy, myofibrillar, 1; scapuloperoneal syndrome, neurogenic, Kaeser type
ELAC2	Combined oxidative phosphorylation deficiency 17
FHL1	Emery-Dreifuss muscular dystrophy 6, X-linked; myopathy, X-linked, with postural muscle atrophy; reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset; reducing body myopathy, X-linked 1b, with late childhood or adult onset; scapuloperoneal myopathy, X-linked dominant

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GENE	DISORDER
FLNC	Cardiomyopathy, familial hypertrophic, 26; cardiomyopathy, familial restrictive 5; myopathy, distal, 4; myopathy, myofibrillar, 5
GAA	Pompe disease; Glycogen storage disease II
GATA4	Atrial septal defect 2; atrioventricular septal defect 4; Tetralogy of Fallot; ventricular septal defect 1
GLA	Fabry disease; Fabry disease, cardiac variant
HRAS	Bladder cancer, somatic; congenital myopathy with excess of muscle spindles; Costello syndrome; nevus sebaceous or woolly hair nevus, somatic; Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic; Spitz nevus or nevus spilus, somatic; thyroid carcinoma, follicular, somatic
JPH2	Cardiomyopathy, dilated, 2E; cardiomyopathy, hypertrophic, 17
KRAS	Arteriovenous malformation of the brain, somatic; bladder cancer, somatic; breast cancer, somatic; cardiofaciocutaneous syndrome 2; gastric cancer, somatic; leukemia, acute myeloid, somatic; lung cancer, somatic; Noonan syndrome 3; oculoectodermal syndrome, somatic; pancreatic carcinoma, somatic; RAS-associated autoimmune leukoproliferative disorder; Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic
LAMP2	Danon disease
LDB3	Cardiomyopathy, dilated, 1C, with or without LVNC; cardiomyopathy, hypertrophic, 24; left ventricular noncompaction 3; myopathy, myofibrillar, 4
MAP2K1	Cardiofaciocutaneous syndrome 3; melorheostosis, isolated, somatic mosaic
MAP2K2	Cardiofaciocutaneous syndrome 4
MTO1	Combined oxidative phosphorylation deficiency 10
MYBPC3	Cardiomyopathy, dilated, 1MM; cardiomyopathy, hypertrophic, 4; left ventricular noncompaction 10

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GENE	DISORDER
MYH6	Atrial septal defect 3; cardiomyopathy, dilated, 1EE; cardiomyopathy, hypertrophic, 14
MYH7	Cardiomyopathy, dilated, 1S; cardiomyopathy, hypertrophic, 1; Laing distal myopathy; left ventricular noncompaction 5; myopathy, myosin storage, autosomal dominant; myopathy, myosin storage, autosomal recessive; scapuloperoneal syndrome, myopathic type
MYL2	Cardiomyopathy, hypertrophic, 10; myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy
MYL3	Cardiomyopathy, hypertrophic, 8
MYLK2	Cardiomyopathy, hypertrophic, 1, digenic
MYOM1	Cardiomyopathy, hypertrophic
MYOZ2	Cardiomyopathy, hypertrophic, 16
MYPN	Cardiomyopathy, dilated, 1KK; cardiomyopathy, familial restrictive, 4; cardiomyopathy, hypertrophic, 22; nemaline myopathy 11, autosomal recessive
NEXN	Cardiomyopathy, dilated, 1CC; cardiomyopathy, hypertrophic, 20

GENE	DISORDER
NF1	Leukemia, juvenile myelomonocytic; neurofibromatosis, familial spinal; neurofibromatosis, type 1; neurofibromatosis-Noonan syndrome; Watson syndrome
NRAS	Colorectal cancer, somatic; epidermal nevus, somatic; melanocytic nevus syndrome, congenital, somatic; neurocutaneous melanosis, somatic; Noonan syndrome 6; Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic; thyroid carcinoma, follicular, somatic
PDLIM3	Cardiomyopathy, dilated; cardiomyopathy, hypertrophic
PLN	Cardiomyopathy, dilated, 1P; cardiomyopathy, hypertrophic, 18
PRKAG2	Cardiomyopathy, hypertrophic 6; glycogen storage disease of heart, lethal congenital; Wolff-Parkinson-White syndrome
PTPN11	LEOPARD syndrome 1; leukemia, juvenile myelomonocytic, somatic; metachondromatosis; Noonan syndrome 1
RAF1	Cardiomyopathy, dilated, 1NN; LEOPARD syndrome 2; Noonan syndrome 5
RASA1	Basal cell carcinoma, somatic; capillary malformation-arteriovenous malformation 1

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GENE	DISORDER
RIT1	Noonan syndrome 8
RRAS	Noonan syndrome
SHOC2	Noonan syndrome-like with loose anagen hair 1
SOS1	Noonan syndrome 4
SOS2	Noonan syndrome 9
SPRED1	Legius syndrome
TCAP	Cardiomyopathy, hypertrophic, 25; muscular dystrophy, limb-girdle, autosomal recessive 7
TNNC1	Cardiomyopathy, dilated, 1Z; cardiomyopathy, hypertrophic, 13
TNNI3	Cardiomyopathy, dilated, 1FF; cardiomyopathy, familial restrictive, 1; cardiomyopathy, hypertrophic, 7
TNNT2	Cardiomyopathy, dilated, 1D; cardiomyopathy, familial restrictive, 3; cardiomyopathy, hypertrophic, 2; left ventricular noncompaction 6
TPM1	Cardiomyopathy, dilated, 1Y; cardiomyopathy, hypertrophic, 3; left ventricular noncompaction 9
TTR	Amyloidosis, hereditary, transthyretin-related; carpal tunnel syndrome, familial
VCL	Cardiomyopathy, dilated, 1W; cardiomyopathy, hypertrophic, 15

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