

別紙

Gene	Phenotype group	Disorder (MIM category)	Inheritance	Variants to report	Triage	Triage rationale (concise)
APC	Cancer	Familial adenomatous polyposis	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
RET	Cancer	Familial medullary thyroid cancer / MEN2	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
BRCA1	Cancer	Hereditary breast/ovarian cancer	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
BRCA2	Cancer	Hereditary breast/ovarian cancer	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
PALB2	Cancer	Hereditary breast cancer	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
SDHD	Cancer	Paraganglioma-pheochromocytoma	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
SDHAF2	Cancer	Paraganglioma-pheochromocytoma	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
SDHC	Cancer	Paraganglioma-pheochromocytoma	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
SDHB	Cancer	Paraganglioma-pheochromocytoma	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
MAX	Cancer	Paraganglioma-pheochromocytoma	AD	All P/LP	Priority 2	介入可能だが浸透率/発症年齢に幅。個別化監視中心
TMEM127	Cancer	Paraganglioma-pheochromocytoma	AD	All P/LP	Priority 2	介入可能だが浸透率/発症年齢に幅。個別化監視中心
BMPR1A	Cancer	Juvenile polyposis syndrome	AD	All P/LP	Priority 2	介入可能だが浸透率/発症年齢に幅。個別化監視中心
SMAD4	Cancer	Juvenile polyposis / HHT overlap	AD	All P/LP	Priority 2	介入可能だが浸透率/発症年齢に幅。個別化監視中心
TP53	Cancer	Li-Fraumeni syndrome	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
MLH1	Cancer	Lynch syndrome	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
MSH2	Cancer	Lynch syndrome	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
MSH6	Cancer	Lynch syndrome	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
PMS2	Cancer	Lynch syndrome	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
MEN1	Cancer	Multiple endocrine neoplasia type 1	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
MUTYH	Cancer	MUTYH-associated polyposis	AR	P/LP (biallelic)	Priority 2	高浸透率・監視/予防/治療が確立。早期介入の利益が大
NF2	Cancer	Neurofibromatosis type 2	AD	All P/LP	Priority 2	常劣; biallelicで高リスク。ヘテロは管理不確実
STK11	Cancer	Peutz-Jeghers syndrome	AD	All P/LP	Priority 1	介入可能だが浸透率/発症年齢に幅。個別化監視中心
PTEN	Cancer	PTEN hamartoma tumor syndrome	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
RB1	Cancer	Retinoblastoma	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
TSC1	Cancer	Tuberous sclerosis complex	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
TSC2	Cancer	Tuberous sclerosis complex	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
VHL	Cancer	von Hippel-Lindau syndrome	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
WT1	Cancer	WT1-related Wilms tumor	AD	All P/LP	Priority 1	高浸透率・監視/予防/治療が確立。早期介入の利益が大
FBN1	Cardiovascular - Aortopathy	Aortopathies	AD	All P/LP	Priority 1	大動脈解離/破裂の急性致死リスク。予防的手術や厳格管理が有効
TGFBR1	Cardiovascular - Aortopathy	Aortopathies	AD	All P/LP	Priority 1	大動脈解離/破裂の急性致死リスク。予防的手術や厳格管理が有効
TGFBR2	Cardiovascular - Aortopathy	Aortopathies	AD	All P/LP	Priority 1	大動脈解離/破裂の急性致死リスク。予防的手術や厳格管理が有効
SMAD3	Cardiovascular - Aortopathy	Aortopathies	AD	All P/LP	Priority 1	大動脈解離/破裂の急性致死リスク。予防的手術や厳格管理が有効
ACTA2	Cardiovascular - Aortopathy	Aortopathies	AD	All P/LP	Priority 1	大動脈解離/破裂の急性致死リスク。予防的手術や厳格管理が有効
MYH11	Cardiovascular - Aortopathy	Aortopathies	AD	All P/LP	Priority 1	大動脈解離/破裂の急性致死リスク。予防的手術や厳格管理が有効
PKP2	Cardiovascular - ARVC	Arrhythmogenic right ventricular cardiomyopathy	AD	All P/LP	Priority 1	致死性不整脈/SCDリスク。ICD等で予防可能
DSP	Cardiovascular - ARVC	Arrhythmogenic right ventricular cardiomyopathy	AD	All P/LP	Priority 1	致死性不整脈/SCDリスク。ICD等で予防可能
DSC2	Cardiovascular - ARVC	Arrhythmogenic right ventricular cardiomyopathy	AD	All P/LP	Priority 1	致死性不整脈/SCDリスク。ICD等で予防可能
TMEM43	Cardiovascular - ARVC	Arrhythmogenic right ventricular cardiomyopathy	AD	All P/LP	Priority 1	致死性不整脈/SCDリスク。ICD等で予防可能
DSG2	Cardiovascular - ARVC	Arrhythmogenic right ventricular cardiomyopathy	AD	All P/LP	Priority 1	致死性不整脈/SCDリスク。ICD等で予防可能
RYR2	Cardiovascular - Channelopathy	Catecholaminergic polymorphic ventricular tachycardia	AD	All P/LP	Priority 1	運動誘発SCD; β遮断薬/ICDが有効
CASQ2	Cardiovascular - Channelopathy	Catecholaminergic polymorphic ventricular tachycardia	AR	P/LP (biallelic)	Priority 1	運動誘発SCD; 介入で低減
TRDN	Cardiovascular - Channelopathy	CPVT/Long QT	AR	P/LP (biallelic)	Priority 1	小児期SCD; 介入必要
TNNT2	Cardiovascular - DCM	Dilated cardiomyopathy	AD	All P/LP	Priority 1	心不全・致死性不整脈; 介入可
LMNA	Cardiovascular - DCM	Dilated cardiomyopathy	AD	All P/LP	Priority 1	心伝導障害/悪性不整脈; ICD等で介入
FLNC	Cardiovascular - DCM/arrhythmia	Dilated/arrhythmogenic cardiomyopathy	AD	All P/LP	Priority 1	SCD高リスク; 介入で予防
TTN	Cardiovascular - DCM	Dilated cardiomyopathy	AD	LoFのみ報告	Priority 1	心不全リスク; 管理で予防 (LoFに限定)
COL3A1	Cardiovascular - Connective tissue	Ehlers-Danlos syndrome, vascular type	AD	All P/LP	Priority 1	動脈破裂/子宮破裂など致死合併症
LDLR	Cardiovascular - Lipid	Familial hypercholesterolemia	AD	All P/LP	Priority 1	早期介入 (高強度脂質低下療法) でイベント抑制
APOB	Cardiovascular - Lipid	Familial hypercholesterolemia	AD	All P/LP	Priority 1	早期介入 (高強度脂質低下療法) でイベント抑制
PCSK9	Cardiovascular - Lipid	Familial hypercholesterolemia	AD	All P/LP	Priority 1	早期介入 (高強度脂質低下療法) でイベント抑制
MYH7	Cardiovascular - HCM	Hypertrophic cardiomyopathy	AD	All P/LP	Priority 1	SCD/心不全リスク; 監視・薬物/デバイス介入が有効
MYBPC3	Cardiovascular - HCM	Hypertrophic cardiomyopathy	AD	All P/LP	Priority 1	SCD/心不全リスク; 監視・薬物/デバイス介入が有効
TNNI3	Cardiovascular - HCM	Hypertrophic cardiomyopathy	AD	All P/LP	Priority 1	SCD/心不全リスク; 監視・薬物/デバイス介入が有効
TPM1	Cardiovascular - HCM	Hypertrophic cardiomyopathy	AD	All P/LP	Priority 1	SCD/心不全リスク; 監視・薬物/デバイス介入が有効
MYL3	Cardiovascular - HCM	Hypertrophic cardiomyopathy	AD	All P/LP	Priority 1	SCD/心不全リスク; 監視・薬物/デバイス介入が有効
ACTC1	Cardiovascular - HCM	Hypertrophic cardiomyopathy	AD	All P/LP	Priority 1	SCD/心不全リスク; 監視・薬物/デバイス介入が有効
PRKAG2	Cardiovascular - HCM	Hypertrophic cardiomyopathy	AD	All P/LP	Priority 1	SCD/心不全リスク; 監視・薬物/デバイス介入が有効
MYL2	Cardiovascular - HCM	Hypertrophic cardiomyopathy	AD	All P/LP	Priority 1	SCD/心不全リスク; 監視・薬物/デバイス介入が有効
KCNQ1	Cardiovascular - Channelopathy	Long QT (type 1)	AD	All P/LP	Priority 1	SCD予防が可能 (β遮断薬等)
KCNH2	Cardiovascular - Channelopathy	Long QT (type 2)	AD	All P/LP	Priority 1	SCD予防が可能 (β遮断薬等)
SCN5A	Cardiovascular - Channelopathy	LQT3/Brugada	AD	All P/LP	Priority 1	致死性不整脈; 介入可
BTD	Inborn errors of metabolism	Biotinidase deficiency	AR	P/LP (biallelic)	Priority 1	高効果の補充療法 (ビオチン)
GLA	Inborn errors of metabolism / Cardiovascular	Fabry disease	XL	全ヘミ/ヘテロ/ホモのP/LP	Priority 1	ERT等で進行抑制。脳腎/心疾患リスク低減
OTC	Inborn errors of metabolism	Ornithine transcarbamylase deficiency	XL	全ヘミ/ヘテロ/ホモのP/LP	Priority 1	高アンモニア血症致死回避。食事/薬物療法
GAA	Inborn errors of metabolism	Pompe disease	AR	P/LP (biallelic)	Priority 1	ERT有効。早期介入で予後改善
HFE	Miscellaneous	Hereditary hemochromatosis (p.Cys282Tyr homozygotes only)	AR	C282Yホモ接合のみ	Priority 2	有効な瀉血治療。男性で浸透率可変
ACVRL1	Miscellaneous - Vascular	Hereditary hemorrhagic telangiectasia	AD	All P/LP	Priority 2	出血/AVMの監視・塞栓術等で管理
ENG	Miscellaneous - Vascular	Hereditary hemorrhagic telangiectasia	AD	All P/LP	Priority 2	同上 (HHT)
RYR1	Miscellaneous - Pharmacogenetics/Anesthesia	Malignant hyperthermia	AD	All P/LP	Priority 1	全身麻酔誘発の致死性合併症を予防可能 (ダントロレン等)
CACNA1S	Miscellaneous - Pharmacogenetics/Anesthesia	Malignant hyperthermia	AD	All P/LP	Priority 1	同上 (麻酔関連の即時回避可能性)
HNF1A	Miscellaneous - Endocrine	Maturity-onset diabetes of the young (MODY3)	AD	All P/LP	Priority 2	治療方針変更 (SU感受性)・家族検査で利点
RPE65	Miscellaneous - Ophthalmology	RPE65-related retinopathy	AR	P/LP (biallelic)	Priority 2	遺伝子治療対象だが施設限定・適応選別が必要
ATP7B	Miscellaneous - Hepatic	Wilson disease	AR	P/LP (biallelic)	Priority 1	キレート/亜鉛治療で予防可能