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Life Science & System's
Finggal Link

Invitae遺伝学的検査項目一覧表

フィンガルリンク株式会社

【目次】

【保険診療受託測定項目】

TestCode	疾患名	項目名	Gene List	希望納入価
FL3001	栄養障害型 表皮水疱症	Epidermolysis Bullosa (栄養障害型表皮水疱症)	COL7A1	¥80,000
FL3002	先天性QT延長 症候群	Long QT Syndrome (先天性QT延長症候群)	CACNA1C, CALM1, CALM2, CALM3, KCNE1, KCNH2, KCNJ2, KCNQ1, SCN5A, TRDN, AKAP9, ANK2, CAV3, KCNE2, KCNJ5, SNTA1, SCN4B	¥80,000
FL3003	神経有棘 赤血球症	Acanthocytosis (有棘赤血球舞踏病)	VPS13A	¥80,000
FL3004	神経有棘 赤血球症	McLeod syndrome (Mcleod症候群)	XK	¥80,000
FL3005	先天性筋無力 症候群	Congenital Myasthenic Syndromes (先天性筋無力症候群)	CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, AGRN, LRP4, LABM2, MUSK, RAPSN, DOK7, CHAT, SCN4A, GFPT1, DPAGT1, ALG2, ALG14, PLEC, PREPL	¥80,000
FL3006	ペリー症候群	Perry syndrome (ペリー症候群)	DCTN1	¥80,000
FL3007	クルーゾン 症候群	Crouzon syndrome (クルーゾン症候群)	FGFR2, FGFR3	¥80,000
FL3008	ファイファー 症候群	Pfeiffer syndrome (ファイファー症候群)	FGFR1, FGFR2	¥80,000
FL3009	アントレー・ ビクスラー 症候群	Antley-Bixler syndrome (アントレー・ビクスラー症候群)	FGFR2, POR	¥80,000
FL3010	タンジール病	Tangier disease (タンジール病)	ABCA1	¥80,000
FL3011	先天性赤血球 形成異常性貧血	Congenital Dyserythropoietic Anemia (CDA) (先天性赤血球形成異常性貧血)	CDAN1, SEC23B, KIF23, KLF1, GATA1	¥80,000
FL3012	若年発症型 両側性感音難聴	Sensorineural hearing loss (SNHL) (若年発症型両側性感音難聴)	ACTG1,CDH23, COCH, KCNQ4, TECTA, TMPRSS3, WFS1	¥80,000
FL3013	マルファン 症候群	Marfan Syndrome (マルファン症候群)	FBN1, TGFBR1, TGFBR2, SMAD3, TGFB2, TGFB3	¥80,000
FL3014	血管型エーラス ダンロス症候群	Ehlers-Danlos Syndrome (エーラスダンロス症候群 （血管型、古典型、関節型、後側彎型）)	COL5A1, COL5A2, COL3A1, PLOD1, ADAMTS2, CHST14, FKBP14, ATP7A, B3GALT6, B4GALT7, COL12A1. COL1A1, COL1A2, CRTAP, FLNA, P3H1, SLC39A13	¥80,000

TestCode	疾患名	項目名	Gene List	希望納入価
FL3015	遺伝性自己炎症疾患	Hereditary autoinflammatory disease (遺伝性自己炎症疾患)	NLRC4, TREX1, RNASEH2B, RNASEH2C, RNASEH2A, SAMHD1, ADAR, IFIH1, ADA2, TNFAIP3, TREX1	¥80,000
FL3016	エプスタイン症候群	Epstein syndrome (エプスタイン症候群)	MYH9	¥80,000
FL3017	ドラベ症候群	Dravet syndrome (ドラベ症候群)	SCN1A, SCN1B, SCN2A, GABRG2	¥80,000
FL3018	コフィン・シリス症候群	Coffin-Siris syndrome (コフィン・シリス症候群(CSS))	ARID1A, ARID1B, SMARCB1, SMARCA4, SMARCE1, PHF6, SOX11	¥80,000
FL3019	歌舞伎症候群	Kabuki Syndrome (歌舞伎症候群(KS))	KMT2D, KDM6A	¥80,000
FL3020	肺胞蛋白症（自己免疫性又は先天性）	Pulmonary alveolar proteinosis (肺胞蛋白症（自己免疫性又は先天性)	CSF2RA, SFTPB, SFTPC, ABCA3, CSF2RB, NKX2-1, FOXF1, GATA2, OAS1, FARSB, TBX4	¥80,000
FL3021	ヌーナン症候群	Noonan Syndrome (ヌーナン症候群)	PTPN11,SOS1,RAF1,RIT1,KRAS,NRAS,SHOC2,CBL,BRAF, HRAS,MAP2K1,MAP2K2	¥80,000
FL3022	骨形成不全症	Osteogenesis Imperfecta and Bone Fragility (骨形成不全症)	BMP1,COL1A1,COL1A2,CRTAP,FKBP10,IFITM5,P3H1,PPIB,SERPINF1, SERPINH1,SP7,TMEM38B,WNT1, SPARC, FAM46A,MBTPS2,MESDC2	¥80,000
FL3023	非典型溶血性尿毒症症候群	Atypical Hemolytic Uremic Syndrome (非典型溶血性尿毒症症候群)	CFH, CFI, CD46, C3, CFB, THBD, DGKE	¥80,000
FL3024	アルポート症候群	Alport Syndrome (アルポート症候群)	COL4A5, COL4A3, COL4A4	¥80,000
FL3025	ファンコニ貧血	Fanconi anemia (ファンコニ貧血)	FANCA , FANCB , FANCC , BRCA2 , FANCD2, FANCE, FANCF, FANCG, FANCI, BRIP1, FANCL, FANCM , PALB2, RAD51C, SLX4, XPF, RAD51, BRCA1, UBE2T	¥80,000
FL3026	遺伝性鉄芽球性貧血	Sideroblastic anemia (遺伝性鉄芽球性貧血)	ALAS2, SLC25A38, PUS1, ABCB7, GLRX5, SLC19A2	¥80,000
FL3027	アラジール症候群	Alagille Syndrome (アラジール症候群)	JAG1, NOTCH2	¥80,000
FL3028	ルビンシュタイン・テイビ症候群	Rubinstein-Taybi Syndrome (ルビンシュタイン・テイビ症候群)	CREBBP, EP300	¥80,000
FL3029	外胚葉形成不全免疫不全症	PID_Ectodermal dysplasia (外胚葉形成不全)	NFKBIA, IKBKB, ORAI1	¥80,000

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FL3030	原発性免疫不全症候群	PID_Familial hemophagocytic lymphocytosis (家族性血球貪食性リンパ組織球症)	PRF1, UNC13D, STX11, STXBP2, SLC7A7, LYST, RAB27A, AP3B1, AP3D1, SH2D1A, XIAP	¥80,000
FL3031	原発性免疫不全症候群	PID_Autoimmune lymphoproliferative syndrome (自己免疫性リンパ増殖症候群)	FAS, FASLG, CASP8, CASP10, NRAS, KRAS, AIRE, FOXP3, IL2RA, CTLA4, LRBA,STAT3, SH2D1A, IKZF1, PIK3CD, PIK3R1, PRKCD, TNFAIP3	¥80,000
FL3032	原発性免疫不全症候群	PID_Inflammatory bowel disease (炎症性腸疾患)	IL10, IL10RA, IL10RB, NFAT5,TGFB1, RIPK1, FOXP3, IL2RA, CTLA4, LRBA, WAS, XIAP, CYBA, CYBB, NCF2, NCF4, TNFAIP3	¥80,000
FL3033	原発性免疫不全症候群	PID_Chronic granulomatous disease (慢性肉芽腫症)	CYBB,CYBA,NCF2,NCF4,G6PD	¥80,000
FL3034	原発性免疫不全症候群	PID_TLR dysplasia (TLR 異常症)	IRAK4, MYD88, NFKBIA, IKBKB, RPSA, NKX2-5, RBCK1	¥80,000
FL3035	原発性免疫不全症候群	Severe Combined Immunodeficiency (panel1) (重症複合免疫不全症)	IL2RG,JAK3,IL7R,RAG1,RAG2,DCLRE1C,ADA,PNP,ZAP70,LIG4,NHEJ1,TBX1	¥80,000
FL3036	原発性免疫不全症候群	Severe Combined Immunodeficiency (panel2) (重症複合免疫不全症)	AK2,CORO1A,FOXN1,PRKDC,PTPRC,STAT5B,ORAI1,STIM1,MAGT1,RAC2,CHD7,SEMA3E,POLE,ATM,CD3D,CD3E, CD247,LAT	¥80,000
FL3037	原発性免疫不全症候群	PID_MHC deficiency (MHC 欠損症)	TAP1,TAP2,B2M,CIITA,RFXANK,RFX5,RFXAP	¥80,000
FL3038	原発性免疫不全症候群	Unclassifiable immunodeficiency (panel1) (分類不能型免疫不全症)	TNFSF12,TNFRSF13B,TNFRSF13C,CD19,CR2,PLCG2,IKZF1,NFKB1,NFKB2,SEC61A1,IRF2BP2,ATP6AP1,SH3KBP1,ARHGEF1,DNMT3B,ZBTB24,CDCA7,HELLS	¥80,000
FL3039	原発性免疫不全症候群	Unclassifiable immunodeficiency (panel2) (分類不能型免疫不全症)	ICOS,PLCG2,LRBA,CTLA4,IL21R,MALT1,MSN,CARD11,BCL10,ITK,PIK3CD,PIK3R1,NFKB1,NFKB2	¥80,000
FL3040	原発性免疫不全症候群	PID_Neutropenia (panel1) (好中球減少症(panel1))	ELANE,HAX1,WAS,CSF3R,SRP54, CXCR4	¥80,000
FL3041	原発性免疫不全症候群	PID_Neutropenia (panel 2) (好中球減少症(panel2))	GFI1,G6PC3,SLC37A4,TAZ,VPS13B,USB1,JAGN1,CLPB	¥80,000
FL3042	原発性免疫不全症候群	PID_High IgE Syndrome (高 IgE 症候群)	STAT3,TYK2,IL6R,ZNF341,ERBIN,TGFBR1,TGFBR2,SPINK5,PGM3,CARD11,DOCK8	¥80,000
FL3043	原発性免疫不全症候群	PID_Chronic cutaneous mucosal candidiasis (慢性皮膚粘膜カンジダ症)	IL17RA,IL17F,STAT1,TRAF3IP2,RORC,AIRE,STAT3,IL12RB1,IL12B,CARD9	¥80,000
FL3044	原発性免疫不全症候群	PID_B cell defects (B細胞欠損症)	BTK,IGLL1,CD79A,BLNK,PIK3CD,PIK3R1,TCF3,SLC39A7,TRNT1,IKZF1	¥80,000

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FL3045	原発性免疫不全症候群	PID_Prosthetic Deficiency (panel1) (補体欠損症)	C1QA,C1QB,C1QC,C1R,C1S,C2,C3,C5,C6,C7,C8A,C8B,C9,CFB,CFI,CFP,MASP2,MBL2	¥80,000
FL3046	原発性免疫不全症候群	PID_Prosthetic defects(panel2) (補体欠損症)（遺伝性血管性浮腫含む）	SERPING1,F12,ANGPT1,PLG,CD55,CD59	¥80,000
FL3047	原発性免疫不全症候群	PID_Congenital immune def. syndrome 先天性免疫不全症候群(ウイルス易感染性）	STAT1,STAT2,IRF7,IFNAR1,IFIH1,TLR3,TBK1,DBR1,IRF8,MCM4,TMC6,TMC8,CXCR4	¥80,000
FL3048	原発性免疫不全症候群	PID_Mendel's Genotype Mycobacteria Infect (メンデル遺伝型マイコバクテリア易感染症)	IL12RB1, IL12B, IL12RB2, IL23R, IFNGR1, IFNGR2, STAT1, CYBB, IRF8, TYK2, RORC, JAK1, GATA2	¥80,000
FL3049	原発性免疫不全症候群	PID_High IgM Syndrome (高IgM 症候群)	CD40LG, AICDA, CD40, UNG, PIK3CD, PIK3R1, PTEN	¥80,000
FL3050	原発性免疫不全症候群	PID_IPEX syndrome (IPEX 症候群)	FOXP3, IL2RA, IL2RB, CTLA4, LRBA, STAT3, FERMT1,STAT1, STAT5B	¥80,000
FL3051	原発性免疫不全症候群	PID_Wiskott-Aldrich syndrome (ウィスコットアルドリッチ症候群)	WAS, ARPC1B, CDC42, WIPF1	¥80,000
FL3052	原発性免疫不全症候群	PID_Dyskeratosis congenital (先天性角化異常症)	DKC1, TERC, TERT, TINF2, RTEL1, ACD, WRAP53, PARN, CTC1, DCLRE1C	¥80,000
FL3053	原発性免疫不全症候群	PID_Familial dendritic cell deficiency (家族性樹状細胞欠損症)	GATA2, CSF2RA, CSF2RB, IRF7, IRF8	¥80,000
FL3054	原発性免疫不全症候群	PID Immunodeficiency with osteogenesisimperf. (骨形成不全を伴う免疫不全症)	SMARCAL1, RNU4ATAC, EXTL3	¥80,000
FL3055	原発性免疫不全症候群	PID_DNA repair disorders (DNA 修復異常症)	ATM, MRE11, NBN, RAD50, LIG4, NHEJ1, DCLRE1C, PRKDC, DNMT3B, ZBTB24, CDCA7, HELLS, RNF168, MCM4, BLM	¥80,000
FL3056	原発性免疫不全症候群	PID_EB virus-related lymphoproliferative dis (EB ウイルス関連リンパ増殖性疾患)	SH2D1A, XIAP, CD27, RASGRP1, CARMIL2, MAGT1, PRKCD, STK4, ITK, ZAP70, MCM4, PIK3CD, PIK3R1, NFKB1, CTLA4, PRF1, STXBP2, FAS	¥80,000
FL3057	原発性免疫不全症候群	PID_Leukocyte insufficiency (白血球粘着不全症)	ITGB2, SLC35C1, FERMT3, RASGRP2	¥80,000
FL3058	原発性免疫不全症候群	PID_Phagocytic dysfunction (食細胞機能異常症)	RAC2, ACTB, FPR1, CTSC, WDR1, MKL1, CEBPE, G6PD	¥80,000

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FL3059	尿素サイクル異常症	Urea Cycle Disorders_OTC (尿素サイクル異常症)	OTC, NAGS, CPS1, SLC25A15, ARG1, ASS1, ASL	¥80,000
FL3060	フェニルケトン尿症	Urea Cycle Disorders_PKU (フェニルケトン尿症検査)	PAH, GCH1, PCBD1,PTS, QDPR, SPR, DNAJC12	¥80,000
FL3061	メープルシロップ尿症	Urea Cycle Disorders_MSUD (メープルシロップ尿症検査)	BCKDHA, BCKDHB, DBT, DLD	¥80,000
FL3062	ホモシスチン尿症	Urea Cycle Disorders_Homocystinuria (ホモシスチン尿症検査)	CBS, MTRR, MTR, MMACHC, MMADHC, LMBRD1, MTHFR	¥80,000
FL3063	シトルリン血症（Ⅰ型）	Urea Cycle Disorders_Citrullinemia (シトルリン血症（Ⅰ型）検査)	ASS1, SLC25A13, ASL	¥80,000
FL3064	アルギノコハク酸血症	Urea Cycle Disorders_Arginosuccinic acidemia (アルギノコハク酸血症)	ASL	¥80,000
FL3065	メチルマロン酸血症	Urea Cycle Disorders_Methylmalonic acidemia (メチルマロン酸血症検査)	MUT, PCCA, PCCB, ABCD4, HCFC1, LMBRD1, MMAA, MMAB, MMACHC, MMADHC	¥80,000
FL3066	プロピオン酸血症	Urea Cycle Disorders_Propionic acidemia (プロピオン酸血症検査)	MUT, PCCA, PCCB	¥80,000
FL3067	イソ吉草酸血症	Urea Cycle Disorders_Isovaleric acidemia (イソ吉草酸血症検査)	IVD	¥80,000
FL3068	メチルクロトニルグリシン尿症	Urea Cycle Disorders_MGC (メチルクロトニルグリシン尿症検査)	MCCC1, MCCC2	¥80,000
FL3069	HMG血症	Urea Cycle Disorders_HMG-CoA reductase HMG 血症(HMG-CoA リアーゼ欠損症）	HMGCL	¥80,000
FL3070	複合カルボキシラーゼ欠損症	Urea Cycle Disorders_Complex carboxylase def (複合カルボキシラーゼ欠損症)	HLCS, BTD	¥80,000
FL3071	グルタル酸血症（Ⅰ型）	Urea Cycle Disorders_Glutaric acidemia type1 (グルタル酸血症Ⅰ型)	GCDH	¥80,000
FL3072	MCAD欠損症	Urea Cycle Disorders_MCAD (中鎖アシル CoA 脱水素酵素欠損症)	ACADM	¥80,000
FL3073	VLCAD欠損症	Urea Cycle Disorders_VLCAD (極長鎖アシル CoA 脱水素酵素欠損症)	ACADVL	¥80,000

TestCode	疾患名	項目名	Gene List	希望納入価
FL3074	MTP(LCHAD) 欠損症	Urea Cycle Disorders_MTP (三頭酵素欠損症)	HADHA, HADH, ACADVL	¥80,000
FL3075	CPT1欠損症	Urea Cycle Disorders_CPT1 (カルニチンパルミトイルトランスフェラーゼ１欠損症)	CPT1A, CPT2, SLC25A20, SLC22A5	¥80,000
FL3076	先天性銅代謝 異常症	Urea Cycle Disorders_Wilson disease (先天性銅代謝異常症検査)	ATP7A, ATP7B	¥80,000

【保険診療外受託測定項目】

Test Code	疾患群	項目名	Gene List
1101	腫瘍学	Invitaeマルチがんパネル Invitae Multi-Cancer Panel	AIP, ALK, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CHEK2, CTNNA1, DICER1, EGFR, EPCAM, FH, FLCN, GREM1, HOXB13, KIT, LZTR1, MAX, MBD4, MEN1, MET, MITF, MLH1, MSH2, MSH3, MSH6, MUTYH, NF1, NF2, NTHL1, PALB2, PDGFRA, PMS2, POLD1, POLE, POT1, PRKAR1A, PTCH1, PTEN, RAD51C, RAD51D, RB1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL
1102	腫瘍学	Invitae 一般的遺伝性がんパネル Invitae Common Hereditary Cancers Panel	APC, ATM, AXIN2, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, CTNNA1, DICER1, EPCAM, FH, GREM1, HOXB13, KIT, MBD4, MEN1, MLH1, MSH2, MSH3, MSH6, MUTYH, NF1, NTHL1, PALB2, PDGFRA, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, SDHA, SDHB, SDHC, SDHD, SMAD4, SMARCA4, STK11, TP53, TSC1, TSC2, VHL
50001	腫瘍学	Invitae乳がんSTATパネル Invitae Breast Cancer STAT Panel	BRCA1, BRCA2, CDH1, PALB2, PTEN, STK11, TP53
50002	腫瘍学	InvitaeBRCA1およびBRCA2 STATパネル Invitae BRCA1 and BRCA2 STAT Panel	BRCA1, BRCA2
1201	腫瘍学	Invitae 遺伝性乳がんおよび婦人科がんパネル Invitae Hereditary Breast and Gyn Cancers Panel	ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, DICER1, EPCAM, MLH1, MSH2, MSH6, NF1, PALB2, PMS2, PTEN, RAD51C, RAD51D, SMARCA4, STK11, TP53
1204	腫瘍学	Invitae乳がんおよび婦人科がんガイドラインベースパネル Invitae Breast and Gyn Cancers Guidelines-Based Panel	ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, NF1, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53
1206	腫瘍学	Invitae 遺伝性乳がんガイドラインに基づくパネル Invitae Hereditary Breast Cancer Guidelines-Based Panel	ATM, BARD1, BRCA1, BRCA2, CDH1, CHEK2, NF1, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53
1251	腫瘍学	Invitae 遺伝性結腸直腸がんパネル Invitae Hereditary Colorectal Cancer Panel	APC, AXIN2, BLM, BMPR1A, CHEK2, EPCAM, GREM1, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11, TP53
1252	腫瘍学	Invitae結腸直腸癌ガイドラインベースパネル Invitae Colorectal Cancer Guidelines-Based Panel	APC, AXIN2, BMPR1A, CHEK2, EPCAM, GREM1, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11, TP53
1271	腫瘍学	Invitae 遺伝性胃がんパネル Invitae Hereditary Gastric Cancer Panel	APC, ATM, BMPR1A, CDH1, CTNNA1, EPCAM, KIT, MLH1, MSH2, MSH6, NF1, PDGFRA, PMS2, RHBDF2, SDHA, SDHB, SDHC, SDHD, SMAD4, STK11, TP53
1302	腫瘍学	Invitae遺伝性傍神経節腫-褐色細胞腫パネル Invitae Hereditary Paraganglioma-Pheochromocytoma Panel	FH, MAX, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL
1411	腫瘍学	Invitae 遺伝性骨髄異形成症候群/白血病パネル Invitae Myelodysplastic Syndrome/Leukemia Panel	ANKRD26, ATM, BLM, CBL, CEBPA, DDX41, ELANE, EPCAM, ERCC6L2, ETV6, G6PC3, GATA2, GFI1, HAX1, IKZF1, KRAS, MECOM, MLH1, MSH2, MSH6, NBN, NF1, PMS2, PTPN11, RTEL1, RUNX1, SAMD9, SAMD9L, SRP72, TERC, TERT, TP53

1461	腫瘍学	Invitae 遺伝性神経系/脳腫瘍パネル Invitae Hereditary Nervous System/Brain Cancer Panel	AIP, ALK, APC, CDKN1B, CDKN1C, CDKN2A, DICER1, EPCAM, FH, GPC3, HRAS, LZTR1, MAX, MEN1, MLH1, MSH2, MSH6, NBN, NF1, NF2, PHOX2B, PMS2, POT1, PRKAR1A, PTCH1, PTEN, RB1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, SMARCE1, SUFU, TMEM127, TP53, TSC1, TSC2, VHL
1261	腫瘍学	Invitae 遺伝性膵臓がんパネル Invitae Hereditary Pancreatic Cancer Panel	APC, ATM, BRCA1, BRCA2, CDKN2A, EPCAM, MEN1, MLH1, MSH2, MSH6, NF1, PALB2, PMS2, STK11, TP53, TSC1, TSC2, VHL
1362	腫瘍学	Invitae 遺伝性前立腺がんパネル Invitae Hereditary Prostate Cancer Panel	ATM, BRCA1, BRCA2, CHEK2, EPCAM, HOXB13, MLH1, MSH2, MSH6, PALB2, PMS2, TP53
1361	腫瘍学	Invitae 遺伝性腎/尿路がんパネル Invitae Hereditary Renal/Urinary Tract Cancers Panel	BAP1, BLM, BUB1B, CDC73, CDKN1C, CEP57, DICER1, DIS3L2, EPCAM, FH, FLCN, GPC3, MET, MLH1, MSH2, MSH6, PMS2, PTEN, REST, SDHB, SDHC, SMARCA4, SMARCB1, TP53, TRIM28, TRIP13, TSC1, TSC2, VHL, WT1
1511	腫瘍学	Invitae肉腫パネル Invitae Sarcoma Panel	APC, BLM, CDKN1C, DICER1, EPCAM, EXT1, EXT2, FH, HRAS, KIT, MLH1, MSH2, MSH6, NBN, NF1, PDGFRA, PMS2, POT1, PRKAR1A, PTCH1, RB1, RECQL4, SDHA, SDHB, SDHC, SDHD, SUFU, TP53, WRN
1703	腫瘍学	Invitae体質ミスマッチ修復 - 欠損パネル Invitae Constitutional Mismatch Repair-Deficiency Panel	EPCAM, MLH1, MSH2, MSH6, PMS2
1712	腫瘍学	Invitae家族性消化管間質腫瘍症候群パネル Invitae Familial Gastrointestinal Stromal Tumor Syndrome Panel	KIT, NF1, PDGFRA, SDHA, SDHB, SDHC, SDHD
1701	腫瘍学	Invitae BRCA1/2パネル Invitae BRCA1/2 Panel	BRCA1, BRCA2
1707	腫瘍学	Invitae遺伝性びまん性胃がん症候群検査 Invitae Hereditary Diffuse Gastric Cancer Syndrome Test	CDH1
1711	腫瘍学	Invitae 若年性ポリポーシス症候群パネル Invitae Juvenile Polyposis Syndrome Panel	BMPR1A, SMAD4
1742	腫瘍学	Invitae 遺伝性ウィルムス腫瘍パネル Invitae Hereditary Wilms Tumor Panel	BLM, BUB1B, CDC73, CDKN1C, CEP57, DICER1, DIS3L2, GPC3, REST, TP53, TRIM28, TRIP13, WT1
1714	腫瘍学	Invitaeラブドイド腫瘍素因症候群パネル Invitae Rhabdoid Tumor Predisposition Syndrome Panel	SMARCA4, SMARCB1
1303	腫瘍学	Invitae 遺伝性副甲状腺機能亢進症パネル Invitae Hereditary Hyperparathyroidism Panel	AP2S1, CASR, CDC73, CDKN1B, GNA11, MEN1, RET, TRPV6

1101	遺伝性腫瘍	Invitaeマルチがんパネル Invitae Multi-Cancer Panel	AIP, ALK, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CHEK2, CTNNA1, DICER1, EGFR, EPCAM, FH, FLCN, GREM1, HOXB13, KIT, LZTR1, MAX, MBD4, MEN1, MET, MITF, MLH1, MSH2, MSH3, MSH6, MUTYH, NF1, NF2, NTHL1, PALB2, PDGFRA, PMS2, POLD1, POLE, POT1, PRKAR1A, PTCH1, PTEN, RAD51C, RAD51D, RB1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL
1102	遺伝性腫瘍	Invitae 一般的遺伝性がんパネル Invitae Common Hereditary Cancers Panel	APC, ATM, AXIN2, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, CTNNA1, DICER1, EPCAM, FH, GREM1, HOXB13, KIT, MBD4, MEN1, MLH1, MSH2, MSH3, MSH6, MUTYH, NF1, NTHL1, PALB2, PDGFRA, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, SDHA, SDHB, SDHC, SDHD, SMAD4, SMARCA4, STK11, TP53, TSC1, TSC2, VHL
50001	遺伝性腫瘍	Invitae乳がんSTATパネル Invitae Breast Cancer STAT Panel	BRCA1, BRCA2, CDH1, PALB2, PTEN, STK11, TP53
50002	遺伝性腫瘍	InvitaeBRCA1およびBRCA2STATパネル Invitae BRCA1 and BRCA2 STAT Panel	BRCA1, BRCA2
1104	遺伝性腫瘍	Invitae 遺伝性小児固形腫瘍パネル Invitae Hereditary Pediatric Solid Tumors Panel	AIP, ALK, APC, BAP1, BLM, BMPR1A, BUB1B, CDC73, CDK4, CDKN1B, CDKN1C, CDKN2A, CEP57, DICER1, DIS3L2, EPCAM, EXT1, EXT2, FH, GPC3, HRAS, LZTR1, MAX, MEN1, MITF, MLH1, MSH2, MSH6, NBN, NF1, NF2, PHOX2B, PMS2, POT1, PRKAR1A, PTCH1, PTEN, RB1, RECQL4, REST, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TMEM127, TP53, TRIM28, TRIP13, TSC1, TSC2, VHL, WRN, WT1
1201	遺伝性腫瘍	Invitae 遺伝性乳がんおよび婦人科がんパネル Invitae Hereditary Breast and Gyn Cancers Panel	ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, DICER1, EPCAM, MLH1, MSH2, MSH6, NF1, PALB2, PMS2, PTEN, RAD51C, RAD51D, SMARCA4, STK11, TP53
1202	遺伝性腫瘍	Invitae 遺伝性乳がんパネル Invitae Hereditary Breast Cancer Panel	ATM, BARD1, BRCA1, BRCA2, CDH1, CHEK2, NF1, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53
1251	遺伝性腫瘍	Invitae 遺伝性結腸直腸がんパネル Invitae Hereditary Colorectal Cancer Panel	APC, AXIN2, BLM, BMPR1A, CHEK2, EPCAM, GREM1, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11, TP53
1271	遺伝性腫瘍	Invitae 遺伝性胃がんパネル Invitae Hereditary Gastric Cancer Panel	APC, ATM, BMPR1A, CDH1, CTNNA1, EPCAM, KIT, MLH1, MSH2, MSH6, NF1, PDGFRA, PMS2, RHBDF2, SDHA, SDHB, SDHC, SDHD, SMAD4, STK11, TP53
1302	遺伝性腫瘍	Invitae遺伝性傍神経節腫-褐色細胞腫パネル Invitae Hereditary Paraganglioma-Pheochromocytoma Panel	FH, MAX, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL
1303	遺伝性腫瘍	Invitae 遺伝性副甲状腺機能亢進症パネル Invitae Hereditary Hyperparathyroidism Panel	AP2S1, CASR, CDC73, CDKN1B, GNA11, MEN1, RET, TRPV6
1411	遺伝性腫瘍	Invitae 遺伝性骨髄異形成症候群/白血病パネル Invitae Hereditary Myelodysplastic Syndrome/Leukemia Panel	ANKRD26, ATM, BLM, CBL, CEBPA, DDX41, ELANE, EPCAM, ERCC6L2, ETV6, G6PC3, GATA2, GFI1, HAX1, IKZF1, KRAS, MECOM, MLH1, MSH2, MSH6, NBN, NF1, PMS2, PTPN11, RTEL1, RUNX1, SAMD9, SAMD9L, SRP72, TERC, TERT, TP53
1461	遺伝性腫瘍	Invitae 遺伝性神経系/脳腫瘍パネル Invitae Hereditary Nervous System/Brain Cancer Panel	AIP, ALK, APC, CDKN1B, CDKN1C, CDKN2A, DICER1, EPCAM, FH, GPC3, HRAS, LZTR1, MAX, MEN1, MLH1, MSH2, MSH6, NBN, NF1, NF2, PHOX2B, PMS2, POT1, PRKAR1A, PTCH1, PTEN, RB1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, SMARCE1, SUFU, TMEM127, TP53, TSC1, TSC2, VHL
1261	遺伝性腫瘍	Invitae 遺伝性膵臓がんパネル Invitae Hereditary Pancreatic Cancer Panel	APC, ATM, BRCA1, BRCA2, CDKN2A, EPCAM, MEN1, MLH1, MSH2, MSH6, NF1, PALB2, PMS2, STK11, TP53, TSC1, TSC2, VHL

1703	遺伝性腫瘍	Invitae体質ミスマッチ修復 - 欠損パネル Invitae Constitutional Mismatch Repair-Deficiency Panel	EPCAM, MLH1, MSH2, MSH6, PMS2
1707	遺伝性腫瘍	Invitae遺伝性びまん性胃がん症候群検査 Invitae Hereditary Diffuse Gastric Cancer Syndrome Test	CDH1
1362	遺伝性腫瘍	Invitae 遺伝性前立腺がんパネル Invitae Hereditary Prostate Cancer Panel	ATM, BRCA1, BRCA2, CHEK2, EPCAM, HOXB13, MLH1, MSH2, MSH6, PALB2, PMS2, TP53
1361	遺伝性腫瘍	Invitae 遺伝性腎/尿路がんパネル Invitae Hereditary Renal/Urinary Tract Cancers Panel	BAP1, BLM, BUB1B, CDC73, CDKN1C, CEP57, DICER1, DIS3L2, EPCAM, FH, FLCN, GPC3, MET, MLH1, MSH2, MSH6, PMS2, PTEN, REST, SDHB, SDHC, SMARCA4, SMARCB1, TP53, TRIM28, TRIP13, TSC1, TSC2, VHL, WT1
1511	遺伝性腫瘍	Invitae肉腫パネル Invitae Sarcoma Panel	APC, BLM, CDKN1C, DICER1, EPCAM, EXT1, EXT2, FH, HRAS, KIT, MLH1, MSH2, MSH6, NBN, NF1, PDGFRA, PMS2, POT1, PRKAR1A, PTCH1, RB1, RECQL4, SDHA, SDHB, SDHC, SDHD, SUFU, TP53, WRN
1301	遺伝性腫瘍	Invitae 遺伝性甲状腺がんパネル Invitae Hereditary Thyroid Cancer Panel	APC, CHEK2, DICER1, PRKAR1A, PTEN, RET, TP53, WRN
444743	遺伝性腫瘍	Invitae DNA損傷修復パネル Invitae DNA Damage Repair Panel	ARID1A, ATM, ATR, BAP1, BARD1, BLM, BRCA1, BRCA2, BRIP1, CHEK2, EPCAM, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, GEN1, MLH1, MRE11, MSH2, MSH6, NBN, PALB2, PMS2, RAD50, RAD51, RAD51C, RAD51D, SLX4, WRN, XRCC2
1701	遺伝性腫瘍	Invitae BRCA1/2パネル Invitae BRCA1/2 Panel	BRCA1, BRCA2
1712	遺伝性腫瘍	Invitae家族性消化管間質腫瘍症候群パネル Invitae Familial Gastrointestinal Stromal Tumor Syndrome Panel	KIT, NF1, PDGFRA, SDHA, SDHB, SDHC, SDHD
1711	遺伝性腫瘍	Invitae若年性ポリポーシス症候群パネル Invitae Juvenile Polyposis Syndrome Panel	BMPR1A, SMAD4
1714	遺伝性腫瘍	Invitaeラブドイド腫瘍素因症候群パネル Invitae Rhabdoid Tumor Predisposition Syndrome Panel	SMARCA4, SMARCB1
1742	遺伝性腫瘍	Invitae 遺伝性ウィルムス腫瘍パネル Invitae Hereditary Wilms Tumor Panel	BLM, BUB1B, CDC73, CDKN1C, CEP57, DICER1, DIS3L2, GPC3, REST, TP53, TRIM28, TRIP13, WT1
57002	遺伝性腫瘍	Invitae 遺伝性神経内分泌腫瘍および副腎皮質癌パネル Invitae Hereditary Neuroendocrine Tumors and Adrenocortical Carcinoma Panel	APC, CDKN1B, CDKN1C, EPCAM, FH, MAX, MEN1, MLH1, MSH2, MSH6, NF1, PMS2, PRKAR1A, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, TP53, TSC1, TSC2, VHL

【保険診療外受託測定項目】

Test Code	疾患群	項目名	Gene List
2101	心臓疾患	Invitae不整脈と心筋症総合パネル Invitae Arrhythmia and Cardiomyopathy Comprehensive Panel	ABCC9, ACADVL, ACTC1, ACTN2, AGL, ALMS1, ALPK3, BAG3, BRAF, CACNA1C, CACNA1D, CALM1, CALM2, CALM3, CASQ2, CBL, CDH2, CPT2, CRYAB, CSRP3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, ELAC2, EMD, EYA4, FHL1, FKRP, FKTN, FLNC, GAA, GATA4, GATA5, GJA5, GLA, HCN4, HRAS, JUP, KCNE1, KCNH2, KCNJ2, KCNQ1, KRAS, LAMP2, LMNA, LZTR1, MAP2K1, MAP2K2, MRAS, MTO1, MYBPC3, MYH7, MYL2, MYL3, MYL4, MYLK3, NF1, NKX2-5, NRAS, PCCA, PCCB, PKP2, PLN, PPA2, PPCS, PPP1CB, PRKAG2, PTPN11, RAF1, RASA1, RBM20, RIT1, RYR2, SCN5A, SDHA, SGCD, SHOC2, SLC22A5, SOS1, SOS2, SPRED1, TAZ, TBX20, TCAP, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRPM4, TTN, TTR, VCL
2212	心臓疾患	Invitaeブルガダ症候群検査 Invitae Brugada Syndrome Test	SCN5A
2213	心臓疾患	Invitaeカテコラミン誘発多形性心室頻拍パネル Invitae Catecholaminergic Polymorphic Ventricular Tachycardia Panel	CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TRDN
2211	心臓疾患	InvitaeQT延長症候群パネル Invitae Long QT Syndrome Panel	CACNA1C, CALM1, CALM2, CALM3, KCNE1, KCNH2, KCNJ2, KCNQ1, SCN5A, TRDN
2214	心臓疾患	Invitae QT短縮症候群パネル Invitae Short QT Syndrome Panel	KCNH2, KCNJ2, KCNQ1
2263	心臓疾患	Invitae不整脈源性心筋症パネル Invitae Arrhythmogenic Cardiomyopathy Panel	ACTN2, BAG3, CDH2, DES, DSC2, DSG2, DSP, EMD, FLNC, JUP, LMNA, PKP2, PLN, PPA2, PRKAG2, RBM20, RYR2, SCN5A, TMEM43, TNNI3, TNNT2, TTN
2251	心臓疾患	Invitae心筋症総合パネル Invitae Cardiomyopathy Comprehensive Panel	ABCC9, ACADVL, ACTC1, ACTN2, AGL, ALMS1, ALPK3, BAG3, BRAF, CACNA1C, CBL, CPT2, CRYAB, CSRP3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, ELAC2, EMD, EYA4, FHL1, FKRP, FKTN, FLNC, GAA, GLA, HCN4, HRAS, JUP, KRAS, LAMP2, LMNA, LZTR1, MAP2K1, MAP2K2, MRAS, MTO1, MYBPC3, MYH7, MYL2, MYL3, MYLK3, NF1, NRAS, PCCA, PCCB, PKP2, PLN, PPCS, PPP1CB, PRKAG2, PTPN11, RAF1, RASA1, RBM20, RIT1, RYR2, SCN5A, SDHA, SGCD, SHOC2, SLC22A5, SOS1, SOS2, SPRED1, TAZ, TBX20, TCAP, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, TTR, VCL
2262	心臓疾患	Invitae拡張型心筋症パネル Invitae Dilated Cardiomyopathy Panel	ABCC9, ACADVL, ACTC1, ACTN2, ALMS1, ALPK3, BAG3, CPT2, CRYAB, CSRP3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, EMD, EYA4, FKRP, FKTN, FLNC, HCN4, JUP, LAMP2, LMNA, MYBPC3, MYH7, MYLK3, PCCA, PCCB, PKP2, PLN, PPCS, RAF1, RBM20, RYR2, SCN5A, SDHA, SGCD, SLC22A5, TAZ, TBX20, TCAP, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, TTR, VCL
2261	心臓疾患	Invitae肥大型心筋症パネル Invitae Hypertrophic Cardiomyopathy Panel	ACADVL, ACTC1, ACTN2, AGL, ALPK3, BAG3, CACNA1C, CPT2, CSRP3, DES, ELAC2, FHL1, FLNC, GAA, GLA, LAMP2, MTO1, MYBPC3, MYH7, MYL2, MYL3, PLN, PRKAG2, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTR, VCL
5201	心臓疾患	Invitae遺伝性ヘモクロマトーシスパネル Invitae Hereditary Hemochromatosis Panel	FTH1, HAMP, HFE, HJV, SLC40A1, TFR2
2265	心臓疾患	Invitae hATTRアミロイドーシス検査 Invitae Hereditary Transthyretin-mediated amyloidosis (hATTR amyloidosis) Test	TTR
4204	心臓疾患	Invitae先天性心疾患パネル Invitae Congenital Heart Disease Panel	ACTC1, ACVR2B, ALMS1, BCOR, BRAF, CASZ1, CBL, CHD7, CRELD1, ELN, FOXH1, GATA4, GATA5, GATA6, GDF1, GJA1, GPC3, HAND1, HAND2, HRAS, JAG1, KDM6A, KMT2D, KRAS, LEFTY2, MAP2K1, MAP2K2, MED13L, MEIS2, MESP1, MYH6, NFATC1, NKX2-5, NKX2-6, NODAL, NOTCH1, NR2F2, NRAS, NSD1, PLD1, PTPN11, RAF1, RBFOX2, RIT1, ROBO1, SHOC2, SMAD6, SOS1, TAB2, TBX1, TBX20, TBX5, TFAP2B, ZFPM2, ZIC3

2352	心臓疾患	Invitae遺伝性出血性毛細血管拡張症および血管奇形パネル Invitae Hereditary Hemorrhagic Telangiectasia and Vascular Malformations Panel	ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4
2401	心臓疾患	Invitae家族性高コレステロール血症パネル Invitae Familial Hypercholesterolemia Panel	APOB, LDLR, LDLRAP1, PCSK9
2351	心臓疾患	Invitae肺動脈性肺高血圧症パネル Invitae Pulmonary Arterial Hypertension Panel	ACVRL1, AQP1, ATP13A3, BMPR2, CAV1, EIF2AK4, ENG, GDF2, KCNK3, SMAD9, SOX17, TBX4
2301	心臓疾患	Invitae大動脈障害総合パネル Invitae Aortopathy Comprehensive Panel	ACTA2, ADAMTS10, BGN, CBS, COL3A1, COL5A1, COL5A2, EFEMP2, FBN1, FBN2, FLNA, FOXE3, LOX, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2
2313	心臓疾患	Invitaeエーラス-ダンロス症候群パネル Invitae Ehlers-Danlos Syndrome Panel	ADAMTS2, ATP7A, B3GALT6, B4GALT7, CHST14, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, CRTAP, FKBP14, FLNA, P3H1, PLOD1, SLC39A13
2311	心臓疾患	Invitaeロイス-ディーツ症候群パネル Invitae Loeys-Dietz Syndrome Panel	FBN1, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2
2312	心臓疾患	Invitaeマルファン症候群検査 Invitae Marfan Syndrome Test	FBN1
2201	心臓疾患	Invitae不整脈総合パネル Invitae Arrhythmia Comprehensive Panel	ABCC9, ACTN2, BAG3, CACNA1C, CACNA1D, CALM1, CALM2, CALM3, CASQ2, CDH2, DES, DSC2, DSG2, DSP, EMD, FLNC, GATA4, GATA5, GJA5, HCN4, JUP, KCNE1, KCNH2, KCNJ2, KCNQ1, LMNA, MYL4, NKX2-5, PKP2, PLN, PPA2, PRKAG2, RBM20, RYR2, SCN5A, TMEM43, TNNI3, TNNT2, TRDN, TRPM4, TTN
53698	心臓疾患	Invitae包括的な脂質血症パネル Invitae Comprehensive Lipidemia Panel	ABCA1, ABCG5, ABCG8, ANGPTL3, APOA1, APOA5, APOB, APOC2, CETP, CREB3L3, CYP27A1, GPD1, GPIHBP1, LCAT, LDLR, LDLRAP1, LIPA, LIPG, LMF1, LPL, LRP6, MTTP, PCSK9, PNPLA2, SAR1B
1722	皮膚疾患	Invitae基底細胞母斑症候群パネル Invitae Basal Cell Nevus Syndrome Panel	PTCH1, SUFU
1720	皮膚疾患	Invitae BHD症候群検査 Invitae Birt-Hogg-Dubé Syndrome Test	FLCN
1702	皮膚疾患	Invitaeリンチ症候群パネル Invitae Lynch Syndrome panel	EPCAM, MLH1, MSH2, MSH6, PMS2
1713	皮膚疾患	Invitae膵臓がん・悪性黒色腫症候群パネル Invitae Melanoma-Pancreatic Cancer Syndrome Panel	CDK4, CDKN2A
1561	皮膚疾患	Invitae 遺伝性皮膚がんパネル Invitae Hereditary Skin Cancer Panel	BAP1, BLM, BRCA2, CDK4, CDKN2A, EPCAM, MBD4, MITF, MLH1, MSH2, MSH6, PMS2, POT1, PTCH1, PTEN, RB1, SUFU, TP53, WRN
4167	皮膚疾患	Invitae神経線維腫症2型検査 Invitae Neurofibromatosis Type 2 Test	NF2
4168	皮膚疾患	Invitae多発性神経鞘腫症パネル Invitae Schwannomatosis Panel	LZTR1, NF2, SMARCB1
1721	皮膚疾患	Invitae結節性硬化症パネル Invitae Tuberous Sclerosis Complex Panel	TSC1, TSC2
434342	皮膚疾患	InvitaeAdams-オリバー症候群パネル Invitae Adams-Oliver Syndrome Panel	ARHGAP31, DLL4, DOCK6, EOGT, KCTD1, NOTCH1, RBPJ, UBR1
4163	皮膚疾患	Invitae Cardio-Facio-Cutaneous Syndrome Panel	BRAF, KRAS, MAP2K1, MAP2K2, SHOC2, SOS1
434346	皮膚疾患	Invitae先天性魚鱗癬パネル Invitae Congenital Ichthyosis Panel	ABCA12, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1S1, AQP5, CAST, CDSN, CERS3, CLDN1, CYP4F22, EBP, ELOVL1, ELOVL4, GJA1, GJB2, GJB3, GJB4, GJB6, KDSR, KRT1, KRT10, KRT2, KRT9, LIPN, LOR, MBTPS2, NIPAL4, PEX7, PHYH, PNPLA1, POMP, SDR9C7, SERPINB7, SERPINB8, SLC27A4, SNAP29, SPINK5, ST14, STS, SULT2B1, SUMF1, TGM1, VPS33B, ZMPSTE24

5021	皮膚疾患	歯の無歯症パネルの有無にかかわらず、Invitae外胚葉異形成症 Invitae Ectodermal Dysplasia with or without Tooth Agenesis Panel	ANTXR1, APCDD1, ATP7A, AXIN2, BCS1L, BMP4, CDH3, CDSN, CLDN1, CTSC, DSG4, DSP, EDA, EDAR, EDARADD, EGFR, ERCC2, ERCC3, ERCC8, GJA1, GJB2, GJB6, GRHL2, GTF2E2, GTF2H5, HOXC13, HR, IRF6, JUP, KANK2, KDF1, KREMEN1, KRT14, KRT16, KRT17, KRT25, KRT6A, KRT6B, KRT6C, KRT71, KRT74, KRT83, KRT85, LIPH, LPAR6, LRP6, LSS, LTBP3, MBTPS2, MPLKIP, MSX1, NECTIN1, NECTIN4, NFKBIA, PAX9, PKP1, POC1A, PORCN, PTH1R, RIN2, RNF113A, RPL21, SMOC2, SNRPE, SOX18, SPINK5, TFAP2B, TP63, TRPS1, TRPV3, TSPEAR, WNT10A, WNT10B
4165	皮膚疾患	Invitaeレジウス症候群テスト Invitae Legius Syndrome Test	SPRED1
434348	皮膚疾患	Invitae眼皮膚白皮症パネル Invitae Oculocutaneous Albinism Panel	AP3B1, AP3D1, BLOC1S3, BLOC1S6, C10ORF11, DTNBP1, FRMD7, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, MLPH, MYO5A, OCA2, RAB27A, SLC24A5, SLC38A8, SLC45A2, TYR, TYRP1
4735	皮膚疾患	Invitae vanderWoude症候群パネル Invitae van der Woude Syndrome Panel	GRHL3, IRF6
434351	皮膚疾患	Invitae色素性乾皮症パネル Invitae Xeroderma Pigmentosum Panel	DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC
4412	内分泌学	Invitaeアンドロゲン不応症パネル Invitae Androgen Insensitivity Panel	AR, SRD5A2
55007	内分泌学	Invitae性発達障害パネル Invitae Disorders of Sex Development Panel	AMH, AMHR2, ANOS1, AR, ARX, ATRX, B3GLCT, CBX2, CCNQ, CHD7, CKAP2L, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, DHH, DMRT1, ESR2, FRAS1, FREM2, H6PD, HHAT, HOXA13, HSD17B3, HSD3B2, KL, LHCGR, LHX3, LHX4, MAMLD1, MAP3K1, NR0B1, NR5A1, POR, PROP1, PSMC3IP, RSPO1, SOX9, SPECC1L, SRD5A2, SRY, STAR, TNK2, TOE1, TSPYL1, TWIST2, UBR1, WNT4, WNT9B, WT1, WWOX, ZFPM2
4413	内分泌学	女性の性的発達テストのInvitae障害 Invitae Disorders of Female Sex Development Test	SRY
4736	内分泌学	Invitae低ゴナドトロピン性低ゴナドトロピン症パネル Invitae Hypogonadotropic Hypogonadism Panel	ANOS1, AXL, CCDC141, CHD4, CHD7, CYP19A1, DHCR7, DUSP6, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IGSF10, IL17RD, KISS1, KISS1R, KLB, LEP, LEPR, LHB, LMNA, NR0B1, NSMF, PCSK1, PLXNA1, POLR3B, PROK2, PROKR2, PROP1, RELN, RNF216, SEMA3A, SOX10, SOX2, SPRY4, SRA1, SRY, TAC3, TACR3, WDR11
98001	内分泌学	Invitae単一遺伝性肥満パネル Invitae Monogenic Obesity Panel	ADCY3, AFF4, ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BDNF, C8ORF37, CEP164, CEP19, CEP290, CPE, CREBBP, CUL4B, DYRK1B, EP300, FBN3, GNAS, GPR101, IFT172, IFT27, IFT74, KDM6A, KIDINS220, KIF7, KMT2D, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MEGF8, MKKS, MKS1, MRAP2, NLGN2, NPY, NR0B2, NTRK2, PCSK1, PHF6, POMC, PPARG, PRMT7, RAB23, RAI1, RPS6KA3, SCLT1, SDCCAG8, SETD2, SH2B1, SIM1, TRAPPC3, TRAPPC9, TRIM32, TTC8, UCP3, VPS13B, WDPCP
55001	内分泌学	Invitae単発性糖尿病パネル Invitae Monogenic Diabetes Panel	ABCC8, APPL1, BLK, EIF2AK3, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1A, HNF1B, HNF4A, IER3IP1, INS, KCNJ11, KLF11, MNX1, NEUROD1, NEUROG3, NKX2-2, PAX4, PDX1, PPARG, PTF1A, RFX6, SLC19A2, WFS1, ZFP57
98006	内分泌学	Invitae 低血糖パネル Invitae Hypoglycemia Panel	AAAS, ABCC8, ABCD1, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACSF3, ADK, AGL, AKT2, ALDOA, ALDOB, ALG3, ALG6, BCKDHA, BCKDHB, CA5A, CACNA1C, CACNA1D, CDKN1C, COG7, CPT1A, CPT2, CYP7B1, DBH, DBT, DDC, DGUOK, DLD, DMXL2, DOLK, ENO3, ETFA, ETFB, ETFDH, FAH, FBP1, FLAD1, G6PC, GAA, GALE, GALK1, GALT, GBE1, GCK, GH1, GHR, GLUD1, GPC3, GYG1, GYS1, GYS2, HADH, HADHA, HADHB, HESX1, HK1, HMGCL, HMGCS2, HNF1A, HNF4A, HRAS, HSD3B7, INSR, KCNJ11, KDM6A, KMT2D, LAMP2, LDHA, LHX3, MLYCD, MPI, MPV17, NADK2, NNT, NR0B1, NR3C1, NSD1, OPLAH, OTX2, OXCT1, PC, PCK1, PCK2, PCSK1, PDX1, PFKM, PGAM2, PGM1, PHGDH, PHKA1, PHKA2, PHKB, PHKG2, PMM2, POMC, PROP1, PTF1A, PYGL, PYGM, RBCK1, SERAC1, SLC16A1, SLC22A5, SLC25A20, SLC25A32, SLC2A2, SLC37A4, SLC52A1, SLC52A2, SLC52A3, SOX2, SOX3, TAZ, TBX19, TRMT10A, UCP2
72039	内分泌学	Invitae低リン血症パネル Invitae Hypophosphatemia Panel	ALPL, CLCN5, CTNS, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FAM20C, FGF23, FGFR1, GNAS, OCRL, PHEX, SLC34A1, SLC34A3, VDR
72038	内分泌学	InvitaeX-Linked低リン酸血症テスト Invitae X-Linked Hypophosphatemia Test	PHEX
5201	血液疾患	Invitae遺伝性ヘモクロマトーシスパネル Invitae Hereditary Hemochromatosis Panel	FTH1, HAMP, HFE, HJV, SLC40A1, TFR2
5261	血液疾患	Invitaeアンチトロンビン III欠損症検査 Invitae Antithrombin III Deficiency Test	SERPINC1

5251	血液疾患	Invitae遺伝性血栓性素因パネル Invitae Hereditary Thrombophilia Panel	ADAMTS13, F2, F5, F9, FGB, FGG, MPL, PROC, PROS1, SERPINC1, THBD
5262	血液疾患	InvitaeプロテインC欠乏症検査 Invitae Protein C Deficiency Test	PROC
5263	血液疾患	InvitaeプロテインS欠乏症検査 Invitae Protein S Deficiency Test	PROS1
5301	血液疾患	Invitae骨髄不全症候群パネル Invitae Bone Marrow Failure Syndromes Panel	ABCB7, ACD, ADA2, AK2, ALAS2, AP3B1, BRCA2, BRIP1, CD40, CD40LG, CEBPE, CLPB, CSF3R, CTC1, CXCR4, DDX41, DKC1, DNAJC21, EFL1, ELANE, ERCC4, ERCC6L2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, G6PC, G6PC3, GATA1, GATA2, GFI1, HAX1, HTRA2, JAGN1, LYST, MECOM, MPL, MYSM1, NHP2, NOP10, PALB2, PARN, POT1, RAB27A, RBM8A, RECQL4, RMRP, RPL11, RPL15, RPL26, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SLC37A4, SLX4, SMARCD2, SRP54, SRP72, STK4, STN1, TAZ, TCN2, TERC, TERT, TIMM50, TINF2, TP53, UBE2T, USB1, VPS13B, VPS45, WAS, WRAP53
5313	血液疾患	Invitaeダイアモンド・ブラックファン貧血パネル Invitae Diamond-Blackfan Anemia Panel	GATA1, MYSM1, RPL11, RPL15, RPL26, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS29, RPS7
5314	血液疾患	Invitae先天性角化異常症パネル Invitae Dyskeratosis Congenita Panel	ACD, CTC1, DKC1, NHP2, NOP10, PARN, POT1, RTEL1, STN1, TERC, TERT, TINF2, WRAP53
5311	血液疾患	Invitaeファンコニ貧血パネル Invitae Fanconi Anemia Panel	BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, PALB2, SLX4, UBE2T
5314	免疫疾患	Invitae先天性角化異常症パネル Invitae Telomere Biology Disorders Panel	ACD, CTC1, DKC1, NHP2, NOP10, PARN, POT1, RTEL1, STN1, TERC, TERT, TINF2, WRAP53
8111	免疫疾患	Invitae無ガンマグロブリン血症パネル Invitae Agammaglobulinemia Panel	BLM, BLNK, BTK, CCBE1, CD19, CD27, CD79A, CD79B, CD81, CDC42, CDCA7, DNMT3B, FAT4, FNIP1, GATA2, HELLS, ICOSLG, IGLL1, IL2RB, IRF2BP2, IRF4, KMT2A, LIG1, LRRC8A, MOGS, MYSM1, OAS1, PIK3R1, SEC61A1, SH2D1A, SLC39A7, TCF3, TOP2B, TRNT1, XIAP, ZBTB24
8112	免疫疾患	Invitae分類不能型免疫不全症パネル Invitae Common Variable Immunodeficiency Panel	ARHGEF1, ATP6AP1, CD19, CD27, CD81, CR2, CTLA4, CXCR4, DCLRE1C, ICOS, IKZF1, IL21, IL21R, IRF2BP2, JAK3, KDM6A, KMT2D, LRBA, MS4A1, NFKB1, NFKB2, NSMCE3, PIK3CD, PIK3R1, PLCG2, PRKCD, RAC2, RAG1, RAG2, SH3KBP1, STAT3, STXBP2, TNFRSF13B, TNFRSF13C, TNFSF12, VAV1
8113	免疫疾患	Invitae高IgE症候群パネル Invitae Hyper IgE Syndrome Panel	CARD11, DOCK8, DSG1, ERBIN, IL6R, IL6ST, PGM3, SPINK5, STAT3, TYK2, ZNF341
8114	免疫疾患	Invitae高IgM症候群パネル Invitae Hyper IgM Syndrome Panel	AICDA, CD40, CD40LG, PIK3CD, PIK3R1, UNG
8120	免疫疾患	Invitae自己炎症および自己免疫症候群パネル Invitae Autoinflammatory and Autoimmunity Syndromes Panel	ACP5, ADA, ADA2, ADAM17, ADAR, AICDA, AIRE, ANKZF1, AP3B1, ARPC1B, ASAH1, BACH2, BLOC1S6, BTK, C17ORF62, CARD14, CARD8, CASP10, CASP8, CCBE1, CD27, CD3G, CD40, CD40LG, COPA, CR2, CTLA4, CYBA, CYBB, DCLRE1C, DDX58, DEF6, DKC1, DNASE1L3, DNASE2, DOCK8, DSG1, DUOX2, ELANE, FADD, FAS, FASLG, FCHO1, FOXP3, G6PC3, ICOS, IFIH1, IL10, IL10RA, IL10RB, IL1RN, IL21, IL21R, IL2RA, IL2RB, IL2RG, IL36RN, IRF2BP2, ITCH, ITGAM, ITGB2, ITK, JAK1, LIG4, LPIN2, LRBA, LYN, LYST, MAGT1, MEFV, MVK, NCF2, NCF4, NFAT5, NFKB1, NFKB2, NFKBIA, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, OAS1, ORAI1, OTULIN, PEPD, PIK3CD, PIK3R1, PLCG2, PNP, POLA1, POMP, PRF1, PRKCD, PSMA3, PSMB4, PSMB8, PSMG2, PSTPIP1, RAB27A, RAC2, RAG1, RAG2, RASGRP1, RBCK1, RFX5, RFXANK, RFXAP, RIPK1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF31, RTEL1, SAMHD1, SCO2, SH2D1A, SH3BP2, SI, SIAE, SKIV2L, SLC29A3, SLC37A4, SLC7A7, STAT1, STAT3, STAT4, STAT5B, STIM1, STX11, STXBP2, TBX1, TGFB1, TGFBR1, TGFBR2, TMEM173, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF6B, TNFSF12, TOP2B, TPP2, TREX1, TRNT1, TTC37, TTC7A, UNC13D, UNG, WAS, XIAP, ZAP70, ZNF341
4313	免疫疾患	Invitae家族性地中海熱検査 Invitae Familial Mediterranean Fever Test	MEFV
8122	免疫疾患	Invitae単一遺伝性炎症性腸疾患パネル Invitae Monogenic Inflammatory Bowel Disease Panel	ADA, ADAM17, AICDA, ANKZF1, ARPC1B, BACH2, BTK, C17ORF62, CARD8, CARMIL2, CD3G, CD40, CD40LG, CD55, CTLA4, CYBA, CYBB, DCLRE1C, DKC1, DOCK8, DUOX2, FCHO1, FOXP3, G6PC3, ICOS, IL10, IL10RA, IL10RB, IL21, IL2RA, IL2RB, IL2RG, ITGB2, JAK1, LIG4, LRBA, MEFV, MVK, NCF2, NCF4, NFAT5, NLRC4, NOD2, PIK3CD, PIK3R1, PLCG2, POLA1, RAG1, RAG2, RIPK1, RTEL1, SH2D1A, SI, SKIV2L, SLC37A4, STAT1, STAT3, STIM1, STXBP2, TGFB1, TGFBR1, TGFBR2, TTC37, TTC7A, WAS, XIAP, ZAP70, ZNF341

4312	免疫疾患	Invitae自己炎症症候群パネル Invitae Periodic Fever Syndromes Panel	ADA2, ASAH1, ELANE, LPIN2, MEFV, MVK, NLRC4, NLRP12, NLRP3, POMP, PSMA3, PSMB4, PSMB8, PSMG2, PSTPIP1, SCO2, TNFRSF1A, TRNT1
8130	免疫疾患	Invitae包括的なSCIDおよびCIDパネル Invitae Comprehensive Severe Combined Immunodeficiency (SCID) and Combined Immunodeficiency (CID) Panel	ACD, ADA, AK2, ARPC1B, ATM, B2M, BCL10, BCL11B, BLM, CARD11, CARMIL2, CCBE1, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD8A, CDCA7, CHD7, CIITA, CORO1A, CTC1, CTPS1, CXCR4, DCLRE1C, DKC1, DNMT3B, DOCK2, DOCK8, EPG5, ERBIN, ERCC6L2, EXTL3, FAT4, FCHO1, FNIP1, FOXI3, FOXN1, HELLS, ICOS, ICOSLG, IKBKB, IKZF1, IL21, IL21R, IL2RG, IL6R, IL6ST, IL7R, ITK, JAK3, KDM6A, KMT2D, LAT, LCK, LIG1, LIG4, LRBA, MAGT1, MALT1, MAP3K14, MCM4, MSN, MTHFD1, MYSM1, NBN, NFE2L2, NFKBIA, NHEJ1, NHP2, NOP10, NSMCE3, ORAI1, PARN, PAX1, PGM3, PNP, POLD1, POLE, POLE2, PRKDC, PTPRC, RAC2, RAG1, RAG2, RELA, RELB, RFX5, RFXANK, RFXAP, RHOH, RMRP, RNF168, RNF31, RNU4ATAC, RTEL1, SEMA3E, SGPL1, SH2D1A, SKIV2L, SLC46A1, SMARCAL1, SP110, SPINK5, STAT3, STAT5B, STIM1, STK4, STN1, TAP1, TAP2, TAPBP, TBX1, TCN2, TERC, TERT, TFRC, TINF2, TNFRSF4, TP63, TTC37, TTC7A, WAS, WIPF1, ZAP70, ZBTB24, ZNF341
8151	免疫疾患	Invitae自己免疫性リンパ増殖性疾患（ALPS）パネル Invitae Autoimmune Lymphoproliferative Disorders (ALPS) Panel	CASP10, CASP8, CTLA4, FADD, FAS, FASLG, ITK, MAGT1, PIK3CD, PIK3R1, PRKCD, RELA, STAT3
8152	免疫疾患	Invitae遺伝性血球貪食性リンパ組織球症（HLH）障害パネル Invitae Hereditary Hemophagocytic Lymphohistiocytosis (HLH) Disorders Panel	ADA, AP3B1, AP3D1, BLOC1S6, BTK, CD27, GATA2, IL2RA, IL2RG, ITK, LYST, MAGT1, MVK, NLRC4, PNP, PRF1, RAB27A, SH2D1A, SLC7A7, STX11, STXBP2, UNC13D, WAS, XIAP
8160	免疫疾患	Invitae好中球減少症を伴う食作用障害パネル Invitae Phagocytic Disorders Including Neutropenia Panel	ACTB, AK2, AP3B1, AP3D1, BTK, C17ORF62, CD40, CD40LG, CDC42, CEBPE, CLPB, CSF2RA, CSF2RB, CSF3R, CTSC, CXCR2, CXCR4, CYBA, CYBB, DKC1, DNAJC21, EFL1, EIF2AK3, ELANE, FERMT3, FPR1, G6PC, G6PC3, G6PD, GATA2, GFI1, GINS1, HAX1, HTRA2, HYOU1, ITGB2, JAGN1, KAT6A, LAMTOR2, LPIN2, LYST, MKL1, MSN, MTHFD1, NCF2, NCF4, PGM3, PMM2, RAB27A, RAC2, RMRP, SLC35C1, SLC37A4, SMARCD2, SPINK5, SRP54, SRP72, STAT3, STK4, TAZ, TCN2, TERT, TFRC, TIMM50, TNFSF12, USB1, VPS13B, VPS45, WAS, WDR1, WIPF1
55680	免疫疾患	Invitae遺伝性血管浮腫パネル Invitae Hereditary Angioedema Panel	ANGPT1, F12, PLG, SERPING1
55681	免疫疾患	Invitae先天性下痢症パネル Invitae Congenital Diarrhea Panel	ADA, ADAM17, AICDA, ALG6, ANKZF1, ARPC1B, BACH2, BTK, C17ORF62, CARD8, CARMIL2, CD3G, CD40, CD40LG, CD55, CTLA4, CYBA, CYBB, CYP27A1, DCLRE1C, DEF6, DGAT1, DKC1, DOCK8, DUOX2, FCHO1, FOXP3, G6PC3, GUCY2C, ICOS, IL10, IL10RA, IL10RB, IL21, IL2RA, IL2RB, IL2RG, ITGB2, JAK1, LCT, LIG4, LIPA, LRBA, MEFV, MVK, MYO5B, NCF2, NCF4, NEUROG3, NFAT5, NLRC4, NOD2, PIK3CD, PIK3R1, PLCG2, POLA1, RAG1, RAG2, RIPK1, RTEL1, SAR1B, SH2D1A, SI, SKIV2L, SLC26A3, SLC37A4, SLC5A1, SLC9A3, SPINT2, STAT1, STAT3, STIM1, STX3, STXBP2, TGFB1, TGFBR1, TGFBR2, TTC37, TTC7A, UNC45A, WAS, XIAP, ZAP70, ZNF341
8100	免疫疾患	Invitae原発性免疫不全パネル Invitae Primary Immunodeficiency Panel	ACD, ACP5, ACTB, ADA, ADA2, ADAM17, ADAR, ADGRE2, AICDA, AIRE, AK2, ALG6, ALPK1, ANGPT1, ANKZF1, AP3B1, AP3D1, ARHGEF1, ARPC1B, ASAH1, ATM, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BLOC1S3, BLOC1S6, BTK, C17ORF62, C1QA, C1QB, C1QC, C1S, C2, C3, C5, C6, C7, C8A, C8B, C9, CARD11, CARD14, CARD8, CARD9, CARMIL2, CASP10, CASP8, CBL, CCBE1, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD55, CD59, CD79A, CD79B, CD81, CD8A, CDC42, CDCA7, CEBPE, CFB, CFD, CFH, CFI, CFP, CHD7, CIB1, CIITA, CLCN7, CLPB, COL7A1, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR2, CXCR4, CYBA, CYBB, CYP27A1, DBR1, DCLRE1C, DDX58, DEF6, DGAT1, DIAPH1, DKC1, DNAJC21, DNASE1L3, DNASE2, DNMT3B, DOCK2, DOCK8, DSG1, DTNBP1, DUOX2, EFL1, EIF2AK3, ELANE, EPG5, ERBIN, ERCC2, ERCC3, ERCC6L2, EXTL3, FADD, FANCA, FANCB, FANCE, FANCF, FANCI, FANCL, FAS, FASLG, FAT4, FCHO1, FERMT1, FERMT3, FNIP1, FOXI3, FOXN1, FOXP3, FPR1, G6PC, G6PC3, G6PD, GATA1, GATA2, GFI1, GINS1, GTF2E2, GTF2H5, GUCY2C, HAX1, HELLS, HMOX1, HPS1, HPS3, HPS4, HPS5, HPS6, HTRA2, HYOU1, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNGR1, IFNGR2, IGLL1, IKBKB, IKZF1, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL12RB2, IL17F, IL17RA, IL17RC, IL18BP, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7R, IRAK4, IRF2BP2, IRF4, IRF7, IRF8, IRF9, ISG15, ITCH, ITGAM, ITGB2, ITK, JAGN1, JAK1, JAK3, KAT6A, KDM6A, KMT2A, KMT2D, LAMTOR2, LAT, LCK, LCT, LIG1, LIG4, LIPA, LPIN2, LRBA, LRRC8A, LYN, LYST, MAD2L2, MAGT1, MALT1, MAP3K14, MCM4, MEFV, MKL1, MOGS, MPLKIP, MS4A1, MSN, MTHFD1, MVK, MYD88, MYO5B, MYSM1, NBAS, NBN, NCF2, NCF4, NCKAP1L, NCSTN, NEUROG3, NFAT5, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, NSMCE3, OAS1, ORAI1, OSTM1, OTULIN, PARN, PAX1, PEPD, PGM3, PIK3CD, PIK3R1, PLCG2, PLVAP, PMM2, PNLIP, PNP, POLA1, POLD1, POLD2, POLE, POLE2, POLR3A, POLR3F, POMP, PRF1, PRKCD, PRKDC, PSENEN, PSMA3, PSMB4, PSMB8, PSMG2, PSTPIP1, PTPRC, RAB27A, RAC2, RAG1, RAG2, RANBP2, RASGRP1, RBCK1, REL, RELA, RELB, RFWD3, RFX5, RFXANK, RFXAP, RHOH, RIPK1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF113A, RNF168, RNF31, RNU4ATAC, RORC, RPSA, RTEL1, SAMD9, SAMD9L, SAMHD1, SAR1B, SCO2, SEC61A1, SEMA3E, SERPING1, SGPL1, SH2D1A, SH3BP2, SH3KBP1, SI, SIAE, SKIV2L, SLC10A2, SLC26A3, SLC29A3, SLC35C1, SLC37A4, SLC39A7, SLC46A1, SLC51B, SLC5A1, SLC7A7, SLC9A3, SLX4, SMARCAL1, SMARCD2, SNX10, SP110, SPINK5, SPINT2, SPPL2A, SRP54, SRP72, STAT1, STAT2, STAT3, STAT4, STAT5B, STIM1, STK4, STN1, STX11, STX3, STXBP2, TAOK2, TAP1, TAP2, TAPBP, TAZ, TBX1, TCF3, TCIRG1, TCN2, TERC, TERT, TFRC, TGFB1, TGFBR1, TGFBR2, THBD, TICAM1, TIMM50, TINF2, TLR3, TLR7, TMC6, TMC8, TMEM173, TMPRSS15, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TNFRSF6B, TNFRSF9, TNFSF11, TNFSF12, TONSL, TOP2B, TP63, TPP2, TRAF3, TRAF3IP2, TREX1, TRNT1, TTC37, TTC7A, TYK2, UNC13D, UNC45A, UNC93B1, UNG, USB1, VAV1, VPS13B, VPS45, WAS, WDR1, WIPF1, WNT2B, WRAP53, XIAP, ZAP70, ZBTB24, ZCCHC8, ZNF341

6171	代謝/新生児	Invitaeリソソーム蓄積症新生児スクリーニングパネル Invitae Lysosomal Storage Disorders Newborn Screening Panel	CHIT1, GAA, GALC, GBA, GLA, IDS, IDUA, NPC1, NPC2, SMPD1
6102	代謝/新生児	Invitae 代謝新生児スクリーニング確認パネル Invitae Metabolic Newborn Screening Confirmation Panel	AAAS, ABCD1, ABCD3, ABCD4, ACAD8, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACBD5, ACOX1, ACOX2, ACSF3, ADAR, ADK, AHCY, ALDH4A1, ALDH6A1, ALDH7A1, AMACR, AMT, ARG1, ARSA, ASL, ASPA, ASS1, AUH, BCAP31, BCAT2, BCKDHA, BCKDHB, BTD, CBS, CD320, CFTR, CLPB, CPS1, CPT1A, CPT2, DBT, DLD, DNAJC12, DNAJC19, DNM1L, ECHS1, ETFA, ETFB, ETFDH, ETHE1, FAH, FLAD1, FTCD, G6PD, GAA, GALC, GALE, GALK1, GALM, GALT, GAMT, GATM, GCDH, GCGR, GCH1, GLA, GLDC, GNMT, GSS, HADH, HADHA, HADHB, HCFC1, HIBCH, HLCS, HMGCL, HPD, HSD17B10, HSD17B4, HTRA2, IDS, IDUA, IFIH1, IVD, LMBRD1, MAT1A, MCCC1, MCCC2, MCEE, MFF, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MTHFR, MTR, MTRR, MUT, NADK2, NAGS, NR0B1, OAT, OPA3, OTC, PAH, PC, PCBD1, PCCA, PCCB, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGM1, PNPO, PPM1K, PRDX1, PRODH, PROSC, PTS, QDPR, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SCP2, SERAC1, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC25A32, SLC2A1, SLC52A1, SLC52A2, SLC52A3, SLC6A8, SMPD1, SPR, SUCLA2, SUCLG1, TAT, TAZ, TCN2, THAP11, TIMM50, TMEM70, TREX1, ZNF143
98003	代謝/新生児	Invitae 補足代謝新生児スクリーニングパネル Invitae Supplemental Metabolic Newborn Screening Panel	A4GALT, ACAD9, ALDH18A1, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ARCN1, ATP13A2, ATP6AP1, ATP6AP2, ATP6V0A2, ATP6V1A, ATP6V1E1, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, BOLA3, C1GALT1C1, CA5A, CAD, CANT1, CCDC115, CHST14, CHST3, CHST6, CHSY1, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, COG1, COG2, COG4, COG5, COG6, COG7, COG8, COPA, COPB2, CSGALNACT1, CTSD, CYP27A1, D2HGDH, DDOST, DHCR7, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, DSE, EOGT, EXT1, EXT2, EXTL3, FKRP, FKTN, FUK, FUT8, G6PC3, GALNT3, GANAB, GFPT1, GLRX5, GLUL, GM2A, GMPPA, GMPPB, GNE, GNPTAB, GNS, GORAB, GOSR2, GPAA1, HEXA, HEXB, HGSNAT, HMGCS2, IBA57, ISCA2, ISPD, JAGN1, KCTD7, LARGE1, LFNG, LIAS, LIPT1, LIPT2, MAGT1, MAN1B1, MFSD8, MGAT2, MOGS, MPDU1, MPI, NAGLU, NANS, NFU1, NGLY1, NPC1, NPC2, NUS1, OGT, OXCT1, PAPSS2, PGAP1, PGAP2, PGAP3, PGM3, PIGA, PIGB, PIGC, PIGG, PIGL, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGT, PIGU, PIGV, PIGW, PIGY, PMM2, POFUT1, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPT1, PRKCSH, RFT1, RPN2, RXYLT1, SAR1B, SEC23A, SEC23B, SEC24D, SEC63, SGSH, SLC10A7, SLC16A1, SLC26A2, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A8, SLC6A9, SLC7A7, SLC9A7, SRD5A3, SSR3, SSR4, ST3GAL3, ST3GAL5, STT3A, STT3B, TGDS, TMEM165, TMEM199, TRAK1, TRAPPC11, TRAPPC12, TRAPPC2, TRAPPC6B, TRAPPC9, TRIP11, TUSC3, VMA21, VPS13B, XYLT1, XYLT2
6210	代謝/新生児	Invitae 超長鎖脂肪酸増加因子（X連鎖性副腎白質ジストロフィー・ペルオキシダーゼ病含む）検査パネル Invitae Elevated Very Long Chain Fatty Acids Panel (including X-ALD)	AAAS, ABCD1, ABCD3, ACBD5, ACOX1, ACOX2, ADAR, AMACR, ARSA, BCAP31, CLN2 (TPP1), CLN3, DNM1L, GALC, HSD17B4, IFIH1, MFF, NR0B1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PPT1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SCP2, TREX1
6117	代謝/新生児	Invitaeアルギナーゼ欠損症パネル Invitae Elevated Arginine (Arginase deficiency) Panel	ARG1, GCGR
6118	代謝/新生児	Invitaeシトルリン血症パネル Invitae Elevated Citrulline (Citrullinemia) Panel	ASL, ASS1, DLD, PC, SLC25A13
6104	代謝/新生児	Invitae カルニチン回路異常症（C0/C16+C18増加因子）検査パネル Invitae Elevated C0/(C16+C18) Test	CPT1A
6105	代謝/新生児	Invitae プロピオニルカルニチン（C3）増加因子検査パネル Invitae Elevated C3 Panel	ABCD4, ACSF3, ALDH6A1, BTD, CD320, HCFC1, HLCS, LMBRD1, MCEE, MMAA, MMAB, MMACHC, MMADHC, MUT, PCCA, PCCB, PRDX1, TCN2, THAP11, ZNF143
6106	代謝/新生児	Invitae マロニルカルチニン（C3-DC）増加因子検査パネル Invitae Elevated C3-DC Panel	ACSF3, MLYCD
6107	代謝/新生児	Invitae ブチリルカルチニン（C4）増加因子検査パネル Invitae Elevated C4 Panel	ACAD8, ACADS, ETHE1, FTCD
6108	代謝/新生児	Invitae 3-ヒドロキシブチリルカルニチン（C4-OHカルニチン）増加因子検査パネル Invitae Elevated C4-OH Test	HADH, HIBCH

6110	代謝/新生児	Invitae イソバレリルカルニチン（C5）増加因子検査パネル Invitae Elevated C5 Panel	ACADSB, IVD
6112	代謝/新生児	Invitae 3-ヒドロキシイソバレリルカルニチン（C5-OH）増加因子検査パネル Invitae Elevated C5-OH Panel	ACAT1, AUH, BTD, CLPB, DNAJC19, HLCS, HMGCL, HSD17B10, HTRA2, MCCC1, MCCC2, OPA3, SERAC1, TAZ, TIMM50, TMEM70
6114	代謝/新生児	Invitae VLCAD欠損症検査 Invitae Elevated C14 and C14:1 (VLCAD deficiency) Test	ACADVL
6115	代謝/新生児	Invitae 三頭酵素欠損症（C16-OH,C16:1-OH, C18-OH, C18:1-OH増加因子）検査パネル Invitae Elevated C16-OH, C16:1-OH, C18-OH & C18:1-OH Panel	HADHA, HADHB
6116	代謝/新生児	Invitae カルニチン回路異常症（C16, C16:1, C18, C18:1増加）検査パネル Invitae Elevated C16, C16:1, C18, & C18:1 Panel	CPT2, SLC25A20
6124	代謝/新生児	Invitae グリシン増加因子（グリシン脳症含む）検査パネル Invitae Elevated Glycine Panel (including Glycine Encephalopathy)	AMT, BOLA3, GLDC, GLRX5, IBA57, ISCA2, LIAS, LIPT1, LIPT2, NFU1, PNPO, SLC6A9
6119	代謝/新生児	Invitae メープルシロップ尿症（ロイシン増加）検査パネル Invitae Elevated Leucine（MSUD）パネル	BCAT2, BCKDHA, BCKDHB, DBT, DLD, PPM1K
6125	代謝/新生児	Invitae メチオニン増加因子検査パネル Invitae Elevated Methionine Panel	ADK, AHCY, CBS, FAH, GNMT, MAT1A, SLC25A13
6145	代謝/新生児	Invitae 高フェニルアラニン血症検査パネル Invitae Elevated Phenylalanine (Hyperphenylalaninemia) Panel	DNAJC12, GCH1, PAH, PCBD1, PTS, QDPR, SLC25A13, SPR
6146	代謝/新生児	Invitae 高プロリン血症検査パネル Invitae Elevated Proline (Hyperprolinemia) Panel	ALDH4A1, PRODH
6126	代謝/新生児	Invitae チロシン血症検査パネル Invitae Elevated Tyrosine（Tyrosinemia）Panel	FAH, HPD, TAT
6165	代謝/新生児	Invitae脂肪酸酸化欠乏パネル Invitae Fatty Acid Oxidation Defects Panel	ACAD9, ACADM, ACADS, ACADSB, ACADVL, CPT1A, CPT2, ETFA, ETFB, ETFDH, FLAD1, HADH, HADHA, HADHB, HMGCL, HMGCS2, MLYCD, NADK2, SLC22A5, SLC25A20, SLC25A32, SLC52A1, SLC52A2, SLC52A3, TANGO2
6202	代謝/新生児	Invitae銅代謝障害パネル Invitae Copper Metabolism Disorders Panel	AP1S1, ATP6AP1, ATP6AP2, ATP7A, ATP7B, CCDC115, CP, PGM1, SLC33A1, TMEM199
6123	代謝/新生児	Invitae低シトルリンパネル Invitae Low Citrulline Panel	ALDH18A1, CPS1, NAGS, OTC
6127	代謝/新生児	Invitae メチルマロニルカルチニン（C4-DC）増加因子検査パネル Invitae Elevated C4-DC Panel	SUCLA2, SUCLG1

6103	代謝/ 新生児	Invitae低C0検査 Invitae Low C0 Test	SLC22A5
6140	代謝/ 新生児	Invitaeアルカプトン尿症検査 Invitae Alkaptonuria Test	HGD
6142	代謝/ 新生児	Invitaeシスチン尿症パネル Invitae Cystinuria Panel	PREPL, SLC3A1, SLC7A9
6141	代謝/ 新生児	Invitaeメチルマロン酸血症 およびホモシスチン尿症パネル Invitae Methylmalonic Acidemia and Homocystinuria Panel	ABCD4, ACSF3, ADK, AHCY, ALDH6A1, AMN, CBS, CD320, CUBN, GIF, GNMT, HCFC1, LMBRD1, MAT1A, MCEE, MMAA, MMAB, MMACHC, MMADHC, MTHFD1, MTHFR, MTR, MTRR, MUT, PRDX1, SUCLA2, SUCLG1, TCN1, TCN2, THAP11, ZNF143
6156	代謝/ 新生児	Invitae包括的な糖原病パネル Invitae Comprehensive Glycogen Storage Disease Panel	AGL, ALDOA, ENO3, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, LPIN1, PCK1, PCK2, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, POLG, PYGL, PYGM, RBCK1, SLC2A2, SLC37A4
6152	代謝/ 新生児	Invitaeガラクトース血症パネル Invitae Galactosemia Panel	GALE, GALK1, GALM, GALT, PGM1, SLC25A13, SLC2A2
6153	代謝/ 新生児	Invitae グルコース-6-リン酸脱水素酵素（G6PD）欠損症検査 Invitae Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency Test	G6PD
6159	代謝/ 新生児	Invitae 遺伝性フルクトース不耐性試験検査 Invitae Hereditary Fructose Intolerance Test	ALDOB
6160	代謝/ 新生児	Invitae まれな炭水化物障害パネル Invitae Rare Carbohydrate Disorders Panel	FBP1, SLC5A1
6155	代謝/ 新生児	Invitae 先天性グリコシル化異常症パネル Invitae Congenital Disorders of Glycosylation Panel	A4GALT, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ARCN1, ATP6AP1, ATP6AP2, ATP6V0A2, ATP6V1A, ATP6V1E1, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, C1GALT1C1, CAD, CANT1, CCDC115, CHST14, CHST3, CHST6, CHSY1, COG1, COG2, COG4, COG5, COG6, COG7, COG8, COPA, COPB2, CSGALNACT1, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, DSE, EOGT, EXT1, EXT2, EXTL3, FKRP, FKTN, FUK, FUT8, G6PC3, GALNT3, GANAB, GFPT1, GMPPA, GMPPB, GNE, GNPTAB, GORAB, GOSR2, GPAA1, ISPD, JAGN1, LARGE1, LFNG, MAGT1, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NANS, NGLY1, NUS1, OGT, PAPSS2, PGAP1, PGAP2, PGAP3, PGM1, PGM3, PIGA, PIGB, PIGC, PIGG, PIGL, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGT, PIGU, PIGV, PIGW, PIGY, PMM2, POFUT1, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PRKCSH, RFT1, RPN2, RXYLT1, SAR1B, SEC23A, SEC23B, SEC24D, SEC63, SLC10A7, SLC26A2, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A8, SLC9A7, SRD5A3, SSR3, SSR4, ST3GAL3, ST3GAL5, STT3A, STT3B, TGDS, TMEM165, TMEM199, TRAK1, TRAPPC11, TRAPPC12, TRAPPC2, TRAPPC6B, TRAPPC9, TRIP11, TUSC3, VMA21, VPS13B, XYLT1, XYLT2
6169	代謝/ 新生児	Invitaeケトン症パネル Invitae Ketolysis Disorders Panel	ACAT1, OXCT1, SLC16A1
6166	代謝/ 新生児	Invitae MCAD欠損症検査 Invitae Elevated C6, C8 and C10 (MCAD deficiency) Test	ACADM
6197	代謝/ 新生児	Invitae イソブチリル・イソバレリルカルニチン（C4･C5）増加因子（アシルCoA脱水素酵素欠損症）検査パネル Invitae Elevated C4 and C5 (Multiple Acyl-CoA Dehydrogenase deficiency) Panel	ETFA, ETFB, ETFDH, ETHE1, FLAD1, SLC25A32, SLC52A1, SLC52A2, SLC52A3
3406	代謝/ 新生児	脳内鉄沈着を伴う神経変性パネル Invitae Neurodegeneration with Brain Iron Accumulation Panel	AP4M1, ATP13A2, C19ORF12, COASY, CP, CRAT, DCAF17, FA2H, FTL, FUCA1, GJA1, GTPBP2, KIF1A, PANK2, PLA2G6, REPS1, SCP2, SLC25A42, SQSTM1, WDR45

6170	代謝/新生児	Invitae包括的なリソソーム蓄積症パネル Invitae Comprehensive Lysosomal Storage Disorders Panel	AGA, ARSA, ARSB, ASAH1, ASPA, ATP13A2, CHIT1, CLCN7, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CTNS, CTSA, CTSD, CTSK, FUCA1, GAA, GALC, GALNS, GBA, GLA, GLB1, GM2A, GNE, GNPTAB, GNPTG, GNS, GUSB, HEXA, HEXB, HGSNAT, HYAL1, IDS, IDUA, KCTD7, LAMP2, LIPA, LYST, MAN2B1, MANBA, MCOLN1, MFSD8, MMP14, NAGA, NAGLU, NEU1, NPC1, NPC2, PPT1, PSAP, SCARB2, SGSH, SLC17A5, SMPD1, SUMF1, VPS33A
6172	代謝/新生児	Invitaeシスチノーシス検査 Invitae Cystinosis Test	CTNS
6179	代謝/新生児	InvitaeFarber脂肪性肉芽腫症検査 Invitae Farber Lipogranulomatosis Test	ASAH1
2266	代謝/新生児	Invitaeファブリー病検査 Invitae Fabry Disease Test	GLA
6180	代謝/新生児	InvitaeGM2ガングリオシドーシスパネル Invitae GM2 Gangliosidosis Panel	GM2A, HEXA, HEXB
6173	代謝/新生児	Invitaeクラッベ病検査 Invitae Krabbe Disease Test	GALC
6181	代謝/新生児	Invitaeラシソゾーム酸性リパーゼ欠損症検査 Invitae Lysosomal Acid Lipase Deficiency Test	LIPA
6174	代謝/新生児	Invitae異染性白質ジストロフィーパネル Invitae Metachromatic Leukodystrophy Panel	ARSA, PSAP, SUMF1
6185	代謝/新生児	Invitae ムコ多糖症Plus検査パネル（ムコ多糖症、ムコリピドーシス、オリゴ糖症） Invitae Mucopolysaccharidoses Plus (MPS+) Panel	AGA, ARSB, CTSA, CTSK, FUCA1, GALNS, GLB1, GNE, GNPTAB, GNPTG, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, MAN2B1, MANBA, MBTPS1, MCOLN1, NAGA, NAGLU, NEU1, SGSH, SLC17A5, SUMF1, VPS33A
3405	代謝/新生児	Invitae神経セロイドリポフスチン症パネル Invitae Neuronal Ceroid Lipofuscinoses Panel	ATP13A2, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CTSD, KCTD7, MFSD8, PPT1
6190	代謝/新生児	Invitaeニーマンピック病タイプA型およびB型検査 Invitae Niemann-Pick Disease Types A and B Panel	SMPD1
6176	代謝/新生児	Invitaeニーマンピック病C型パネル Invitae Niemann-Pick Disease Type C Panel	NPC1, NPC2
6177	代謝/新生児	Invitaeポンペ病検査 Invitae Pompe Disease Test	GAA
6178	代謝/新生児	Invitaeサンドホフ病検査 Invitae Sandhoff Disease Test	HEXB
4719	代謝/新生児	Invitaeテイ・サックス病検査 Invitae Tay-Sachs Disease Test	HEXA
6183	代謝/新生児	Invitaeウィルソン病検査 Invitae Wilson Disease Test	ATP7B

6192	代謝/新生児	Invitae3-メチルクロトニルCoAカルボキシラーゼパネル Invitae 3-Methylcrotonyl CoA Carboxylase Panel	MCCC1, MCCC2
6194	代謝/新生児	Invitaeビオチニダーゼ欠損症検査 Invitae Biotinidase Deficiency Test	BTD
6195	代謝/新生児	Invitaeグルタル酸尿症I型検査 Invitae Elevated C5-DC (Glutaric Aciduria Type I) Test	GCDH
6191	代謝/新生児	Invitae有機酸血症パネル Invitae Organic Acidemias Panel	ABCD4, ACAD8, ACADSB, ACAT1, ACSF3, ADK, AGK, AHCY, ALDH6A1, AMN, ASPA, ATP5D, AUH, BCKDHA, BCKDHB, BOLA3, BTD, C19ORF70, CBS, CD320, CLPB, CPS1, CUBN, D2HGDH, DBT, DHTKD1, DLD, DNAJC19, ECHS1, ETFA, ETFB, ETFDH, ETHE1, FBP1, FH, FLAD1, FTCD, GCDH, GIF, GLRX5, GLYCTK, GNMT, GSS, HCFC1, HIBCH, HLCS, HMGCL, HSD17B10, HTRA2, IBA57, IDH2, ISCA2, IVD, L2HGDH, LIAS, LIPT1, LIPT2, LMBRD1, MAT1A, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MTHFR, MTR, MTRR, MUT, NFU1, OGDH, OPA3, OPLAH, OXCT1, PCCA, PCCB, PCK1, POLG, PPM1K, PRDX1, SERAC1, SLC13A3, SLC13A5, SLC25A1, SLC25A19, SLC25A32, SLC52A1, SLC52A2, SLC52A3, SUCLA2, SUCLG1, SUGCT, TAZ, TCN1, TCN2, THAP11, TIMM50, TMEM70, ZNF143
6199	代謝/新生児	Invitaeプロピオン酸血症パネル Invitae Propionic Acidemia Panel	PCCA, PCCB
6225	代謝/新生児	Invitaeオルニチントランスカルバミラーゼ（OTC）欠損症検査 Invitae Ornithine Transcarbamylase (OTC) Deficiency Test	OTC
6212	代謝/新生児	Invitae尿素サイクル異常症パネル Invitae Urea Cycle Disorders Panel	ALDH18A1, ARG1, ASL, ASS1, CPS1, NAGS, OAT, OTC, SLC25A13, SLC25A15
6162	代謝/新生児	Invitae脳クレアチン欠乏パネル Invitae Cerebral Creatine Deficiency Panel	GAMT, GATM, SLC6A8
6161	代謝/新生児	Invitae脳腱黄色腫症検査 Invitae Cerebrotendinous Xanthomatosis Test	CYP27A1
6204	代謝/新生児	Invitae驚愕病パネル Invitae Hereditary Hyperekplexia Panel	ARHGEF9, ASNS, ATAD1, CLPB, CTNNB1, GLRA1, GLRB, GPHN, RPS6KA3, SCN8A, SLC6A5, SLC6A9, TRAK1
6203	代謝/新生児	Invitae神経伝達物質障害パネル Invitae Neurotransmitter Disorders Panel	ABAT, ALDH5A1, ALDH7A1, AMT, ARHGEF9, ASNS, ATAD1, DBH, DDC, DHFR, DNAJC12, GABBR2, GABRA1, GABRA2, GABRB1, GABRB3, GABRG2, GAD1, GCH1, GLDC, GLRA1, GLRB, GOT2, GPHN, GRIN2B, GRIN2D, MAOA, PCBD1, PHGDH, PNPO, PROSC, PSAT1, PSPH, PTS, QDPR, SLC18A2, SLC1A2, SLC1A4, SLC25A22, SLC6A1, SLC6A3, SLC6A5, SLC6A9, SPR, TH
6222	代謝/新生児	Invitae治療可能な神経代謝異常パネル Invitae Treatable Neurometabolic Disorders Panel	ABCD1, ABCD4, ACAT1, AGA, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, AMN, AMT, APTX, ARG1, ARHGEF9, ARSA, ASAH1, ASL, ASNS, ASPA, ASS1, ATAD1, ATP7A, ATP7B, AUH, BCKDHA, BCKDHB, BSCL2, BSND, BTD, CA5A, CAD, CASR, CBS, CD320, CLCNKB, CLDN16, CLDN19, CLN2 (TPP1), CLN3, CLN5, CLN6, CNNM2, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, CP, CPOX, CPS1, CUBN, CYP27A1, DBT, DDC, DHCR7, DHFR, DLAT, DLD, DNAJC12, EGF, ETFA, ETFB, ETFDH, ETHE1, FAM111A, FOLR1, FXYD2, GALC, GAMT, GATM, GCDH, GCH1, GCLC, GIF, GLA, GLB1, GLDC, GLRA1, GLRB, GLUD1, GNS, GOT2, GPHN, GSS, GUSB, HCFC1, HEXA, HEXB, HGSNAT, HLCS, HMBS, HMGCL, HMGCS2, HNF1B, HSD17B10, IDS, IDUA, IVD, KCNA1, KCNJ10, LIPA, LMBRD1, MAN2B1, MCCC1, MCCC2, MCEE, MFSD8, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MOCS2, MSMO1, MTHFR, MTR, MTRR, MUT, NAGLU, NAGS, NPC1, NPC2, NT5C3A, OAT, OTC, OXCT1, PAH, PANK2, PCBD1, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PDSS2, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PGM3, PHGDH, PHYH, PNPO, PPM1K, PRDX1, PROSC, PRPS1, PSAT1, PSPH, PTS, QDPR, RAPSN, SCN4A, SGSH, SLC12A1, SLC12A3, SLC13A5, SLC18A2, SLC19A1, SLC19A2, SLC19A3, SLC1A3, SLC25A13, SLC25A15, SLC25A19, SLC2A1, SLC30A10, SLC39A14, SLC39A8, SLC46A1, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SPR, TAT, TCN1, TCN2, TH, TPK1, TRPM6, TTPA
6207	代謝/新生児	Invitae成人レフサム病パネル Invitae Adult Refsum Disease Panel	PEX7, PHYH

6211	代謝/新生児	Invitaeツェルウェガー症候群パネル Invitae Zellweger Spectrum Disorder Panel	ACBD5, ACOX1, AMACR, HSD17B4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7
6213	代謝/新生児	Invitaeプリン代謝異常パネル Invitae Purine Metabolism Disorders Panel	ADA, ADSL, AMPD1, APRT, ATIC, DGUOK, GPHN, HPRT1, IMPDH1, MOCOS, MOCS1, MOCS2, MOCS3, PNP, PRPS1, SUOX, UMOD, XDH
98004	代謝/新生児	Invitaeピルビン酸代謝および関連障害パネル Invitae Pyruvate Metabolism and Related Disorders Panel	BOLA3, DLAT, DLD, ECHS1, FBXL4, FDX2, GLRX5, GOT2, HIBCH, IBA57, ISCA1, ISCA2, ISCU, LIAS, LIPT1, LIPT2, LONP1, LYRM4, MPC1, NFS1, NFU1, NUBPL, PC, PCK1, PDHA1, PDHB, PDHX, PDK3, PDP1, SLC19A2, SLC19A3, SLC25A1, SLC25A19, SUCLA2, SUCLG1, SUCLG2, TAZ, TPK1
55000	代謝/新生児	Invitae α1ーアンチトリプシン欠乏症検査 Invitae Alpha-1 Antitrypsin Deficiency Test	SERPINA1
6226	代謝/新生児	Invitae急性肝性ポルフィリン症パネル Invitae Acute Hepatic Porphyrias Panel	ALAD, CPOX, HMBS, PPOX
6230	代謝/新生児	Invitae高アンモニア血症パネル Invitae Hyperammonemia Panel	ABCD4, ACADM, ACADVL, ALDH18A1, AMT, ARG1, ASL, ASS1, ATP5A1, ATP5D, ATP5E, ATPAF2, BCKDHA, BCKDHB, BTD, CA5A, CPS1, CPT1A, CPT2, CYC1, DBT, DLAT, DLD, ETFA, ETFB, ETFDH, FBXL4, GLDC, GLUD1, GLUL, HADHA, HADHB, HCFC1, HLCS, HMGCL, IVD, LMBRD1, LYRM7, MCCC1, MCCC2, MCEE, MMAA, MMAB, MMACHC, MMADHC, MTR, MTRR, MUT, NAGS, NBAS, NR1H4, OAT, OTC, PC, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PRDX1, RINT1, SERAC1, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC25A42, SLC7A7, TANGO2, TAZ, TMEM70, TUFM, UMPS, UQCRC2, YARS2
6228	代謝/新生児	Invitae精神症状を伴うメンデル遺伝病パネル Invitae Mendelian Disorders with Psychiatric Symptoms Panel	ABCB4, ABCD1, ADSL, ALAD, ALDH5A1, AMACR, AMT, ANK3, AP1S1, ARG1, ARSA, ARX, ASL, ASS1, ATP13A2, ATP1A3, ATP2A2, ATP7B, B4GALNT1, BCKDHA, BCKDHB, BCKDK, C19ORF12, CA5A, CACNA1A, CBS, CHCHD10, CLCN2, CLCN4, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLPX, CNTNAP2, COASY, COL4A1, CP, CPOX, CPS1, CSTB, CTSD, CYP27A1, DARS, DBH, DBT, DCAF17, DDC, DEPDC5, DLD, DNAJC12, DNAJC6, EHMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPM2A, FA2H, FAH, FECH, FIG4, FOLR1, FUCA1, GALC, GAMT, GATM, GCH1, GFAP, GLA, GLB1, GLDC, GM2A, GNS, GRIA3, GSS, HARS, HEXA, HEXB, HGSNAT, HMBS, HMGCL, HPRT1, HTRA1, IQSEC2, KCNT1, KCTD17, MAN2B1, MANBA, MAOA, MECP2, MED12, MFSD8, MMACHC, MMADHC, MSTO1, MTHFR, MTR, MUT, NAGLU, NAGS, NDP, NHLRC1, NPC1, NPC2, OTC, PAH, PAK3, PANK2, PCBD1, PCCA, PCCB, PCDH19, PLA2G6, PLP1, POLG, PPOX, PPT1, PRDX1, PRKAR1A, PRODH, PSAP, PTS, QDPR, REPS1, RPS6KA3, SETX, SGCE, SGSH, SLC12A6, SLC20A2, SLC25A13, SLC25A15, SLC30A10, SLC39A14, SLC52A1, SLC52A2, SLC52A3, SLC6A19, SLC6A3, SLC6A8, SLC7A7, SPART, SPG11, SPR, SUMF1, TBC1D7, TBX1, TH, TIMM8A, TOR1A, TREX1, TRRAP, TTC19, TWNK, TYMP, TYROBP, UBQLN2, UROD, UROS, VPS13A, WDR45, WFS1, YWHAG, ZFYVE26
6229	代謝/新生児	Invitae代謝性非免疫胎児水腫パネル Invitae Metabolic Non-Immune Fetal Hydrops Panel	AHCY, ALG1, ALG12, ALG8, ALG9, ARSB, ASAH1, CTSA, DHCR7, G6PD, GAA, GALC, GALNS, GBE1, GLB1, GLUL, GNPTAB, GUSB, HADH, HADHA, HADHB, IDUA, LIPA, MVK, NEU1, NPC1, NPC2, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHGDH, PIGA, PMM2, PSAT1, SLC17A5, SLC22A5, SLC26A2, SMPD1, SUMF1, TAZ, TRIP11
6227	代謝/新生児	Invitae原発性高シュウ酸尿症パネル Invitae Primary Hyperoxaluria Panel	AGXT, GRHPR, HOGA1
72039	代謝/新生児	Invitae低リン血症パネル Invitae Hypophosphatemia Panel	ALPL, CLCN5, CTNS, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FAM20C, FGF23, FGFR1, GNAS, OCRL, PHEX, SLC34A1, SLC34A3, VDR
55003	代謝/新生児	Invitae包括的なポルフィリン症パネル Invitae Comprehensive Porphyrias Panel	ALAD, ALAS2, CLPX, CPOX, FECH, GATA1, HMBS, PPOX, UROD, UROS

55002	代謝/新生児	Invitae白質ジストロフィーと遺伝性白質脳症パネル Invitae Leukodystrophy and Genetic Leukoencephalopathy Panel	AARS, AARS2, ABAT, ABCA1, ABCD1, ABCD4, ABHD5, ACADS, ACBD5, ACER3, ACO2, ACOX1, ACP5, ACSF3, ACTA2, ACTB, ACY1, ADAR, ADGRG1, ADK, ADNP, ADSL, AGA, AHDC1, AHI1, AIFM1, AIMP1, AIMP2, ALDH18A1, ALDH3A2, ALDH5A1, ALDH6A1, ALG12, ALG13, ALG2, ALG6, ALG9, AMACR, AMPD2, AMT, ANK3, AP1S2, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APC2, APOPT1, ARCN1, ARFGEF2, ARHGAP31, ARHGEF9, ARNT2, ARSA, ARX, ASL, ASNS, ASPA, ASS1, ASXL1, ASXL2, ATP13A2, ATP5A1, ATP6AP2, ATP6V1A, ATP7A, ATP7B, ATP8A2, ATPAF2, ATRN, ATRX, AUH, B3GALNT2, B4GALNT1, BCAP31, BCAT2, BCKDHA, BCKDHB, BCL11B, BCS1L, BICD2, BMP4, BOLA3, BPTF, BRAT1, BTD, C12ORF57, C12ORF65, C19ORF12, C2CD3, CACNA1A, CACNA1E, CARS2, CBS, CC2D2A, CCDC88A, CDC42, CDKL5, CEP290, CHMP1A, CLCN2, CLCN4, CLCN7, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLP1, CLPP, CNNM2, CNOT1, CNTNAP1, CNTNAP2, COA7, COASY, COG7, COL18A1, COL3A1, COL4A1, COL4A2, COLGALT1, COQ2, COQ6, COQ7, COQ8A, COQ9, COX10, COX14, COX15, COX20, COX6B1, COX7B, COX8A, CPLANE1, CPLX1, CPS1, CRAT, CREBBP, CRIPT, CRLF1, CSF1R, CSPP1, CTBP1, CTC1, CTDP1, CTNS, CTSA, CTSD, CUL4B, CYB5R3, CYFIP2, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS, DARS2, DBT, DCAF17, DCX, DDC, DDHD1, DDHD2, DDOST, DDX3X, DEAF1, DEGS1, DGUOK, DHCR24, DHFR, DHX37, DLD, DLL4, DMXL2, DNM1L, DNM2, DOCK6, DOCK7, DOLK, DONSON, DPAGT1, DPM1, DPM2, DPM3, DPYS, DYRK1A, EARS2, EDNRB, EHMT1, EIF2AK1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL1, ELOVL4, ENTPD1, EOGT, EPG5, EPRS, ERCC1, ERCC2, ERCC3, ERCC6, ERCC8, ETFA, ETFB, ETFDH, ETHE1, EXOSC2, EXOSC3, EXOSC8, EXOSC9, FA2H, FAM126A, FAR1, FARS2, FARSB, FASTKD2, FBXL4, FDX2, FGFRL1, FH, FIG4, FKRP, FKTN, FLVCR2, FOLR1, FOXC1, FOXG1, FOXRED1, FUCA1, FUK, GAA, GABBR2, GABRA1, GABRB1, GABRB2, GABRB3, GALC, GALT, GAN, GATAD2B, GCDH, GDAP1, GFAP, GFM1, GFM2, GJA1, GJB1, GJC2, GLA, GLB1, GLDC, GLRX5, GLUL, GLYCTK, GM2A, GMNN, GNAO1, GNS, GOT2, GPHN, GRIN1, GRM7, GTF2H5, GTPBP2, HACE1, HCN1, HEPACAM, HERC1, HEXA, HEXB, HIBCH, HIKESHI, HK1, HLCS, HMGCL, HNRNPU, HSD17B10, HSD17B4, HSPD1, IARS, IARS2, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, INPP5E, IREB2, ISCA1, ISCA2, ITPA, ITPR1, IVD, JAM3, KARS, KAT6B, KATNB1, KCNJ10, KCNJ2, KCNMA1, KCNT1, KCTD7, KDM1A, KIAA0556, KIAA0586, KIAA0753, KIDINS220, KIF1A, KIF1C, KLHL7, KMT2E, L2HGDH, LAGE3, LAMA1, LAMA2, LAMB1, LARGE1, LARS2, LETM1, LIAS, LIPT1, LIPT2, LONP1, LRPPRC, LYRM7, MAG, MAGEL2, MAN2B1, MANBA, MARS2, MAST1, MAT1A, MCCC1, MCCC2, MCOLN1, MDH2, MECP2, MED17, MED25, MEF2C, MFSD2A, MFSD8, MGP, MICU1, MKS1, MLC1, MLYCD, MMACHC, MMADHC, MOCS1, MOCS2, MOGS, MORC2, MPLKIP, MPV17, MPZ, MRPL12, MRPL44, MRPS16, MRPS22, MRPS34, MSTO1, MTFMT, MTHFR, MTHFS, MTO1, MTOR, MTR, MTRR, MUT, NACC1, NADK2, NAGA, NAGLU, NAGS, NANS, NARS2, NAXD, NAXE, NDRG1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB10, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEK1, NFE2L2, NFU1, NGLY1, NKX6-2, NOTCH1, NPC1, NPC2, NPHP1, NRXN1, NSUN3, NT5C2, NTRK2, NUBPL, NUP62, OAT, OCRL, OPA1, OPA3, OSGEP, OTC, PACS1, PAFAH1B1, PAH, PANK2, PARS2, PAX1, PC, PCCA, PCCB, PCDH12, PCGF2, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGK1, PHGDH, PHYH, PIGA, PIGB, PIGG, PIGL, PIGM, PIGN, PIGP, PIGQ, PIGT, PIGU, PIGV, PIK3C2A, PIK3R2, PLA2G6, PLAA, PLEKHG2, PLP1, PMM2, PMP22, PNKP, PNPT1, POLG, POLG2, POLR1A, POLR1C, POLR3A, POLR3B, POMGNT1, POMK, POMT1, POMT2, PPP1R15B, PPP2R1A, PPP3CA, PPT1, PRF1, PRKDC, PRODH, PROSC, PRPS1, PRUNE1, PSAP, PSAT1, PSPH, PTEN, PTPN23, PURA, PUS3, PYCR2, QARS, QRSL1, RAB11B, RAB3GAP1, RAB3GAP2, RARS, RARS2, RBPJ, REPS1, RERE, RHOBTB2, RIN2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RPGRIP1L, RPIA, RPS6KC1, RRM2B, RTTN, RXYLT1, SAMD9L, SAMHD1, SCN3A, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SEPSECS, SERAC1, SETBP1, SGSH, SH3TC2, SHOC2, SHPK, SLC12A2, SLC12A5, SLC12A6, SLC13A3, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC1A2, SLC1A3, SLC1A4, SLC25A1, SLC25A12, SLC25A15, SLC25A19, SLC25A22, SLC25A4, SLC25A42, SLC25A46, SLC2A1, SLC30A10, SLC33A1, SLC35A2, SLC39A14, SLC39A8, SLC46A1, SLC6A19, SLC6A8, SLC6A9, SLC9A1, SLC9A6, SMC1A, SNAP29, SNIP1, SNORD118, SNRPB, SNX14, SOD1, SON, SOX10, SOX2, SPART, SPAST, SPATA5, SPG11, SPG7, SPTAN1, SQSTM1, SRD5A3, SSR4, ST3GAL5, STAG2, STAMBP, STAT1, STAT2, STN1, STRADA, STX11, STXBP1, STXBP2, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYNE1, SYNJ1, TACO1, TAF2, TANGO2, TARS2, TBC1D24, TBCD, TBCE, TBCK, TBX1, TCF4, TCTN2, TIMM50, TIMMDC1, TM4SF20, TMEM106B, TMEM126B, TMEM165, TMEM216, TMEM67, TMEM70, TMTC3, TOE1, TP53RK, TPI1, TPK1, TRAPPC11, TRAPPC9, TREX1, TRMT10A, TRMT5, TSC1, TSC2, TSEN54, TTC19, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUFM, TWNK, TXN2, TYMP, UBA5, UBE2A, UBE3A, UFM1, UNC13D, UPB1, USP7, VARS2, VPS11, VPS13D, VPS33A, WARS2, WDR45, WDR73, WHSC1, WWOX, YARS, YME1L1, ZEB2, ZFYVE26, ZIC1, ZNF335
72038	代謝/新生児	InvitaeX-Linked低リン酸血症テスト Invitae X-Linked Hypophosphatemia Test	PHEX
4305	代謝/新生児	Invitae Achromatopsia Panel	ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H, RGS9, RGS9BP

98002	代謝/ 新生児	Invitae核ミトコンドリア病パネル Invitae Nuclear Mitochondrial Disorders Panel	AARS2, AASS, ABAT, ABCB7, ACACA, ACAD9, ACADM, ACADS, ACADVL, ACAT1, ACO2, ADAR, AFG3L2, AGK, AIFM1, AK2, ALAS2, ALDH3A2, AMPD1, AMT, APOPT1, APTX, ATP5A1, ATP5D, ATP5E, ATP7B, ATPAF2, AUH, BAG3, BCS1L, BOLA3, BTD, C12ORF65, C19ORF12, C19ORF70, C1QBP, CA5A, CARS2, CEP89, CHAT, CHCHD10, CLPB, CLPP, COA3, COA5, COA6, COA7, COASY, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I2, COX6A1, COX6B1, COX7B, COX8A, CPS1, CPT1A, CPT2, CYC1, CYCS, D2HGDH, DARS2, DES, DGUOK, DLAT, DLD, DNA2, DNAJC19, DNM1L, EARS2, ECHS1, ELAC2, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBXL4, FDX2, FH, FLAD1, FOXRED1, GAMT, GARS, GATM, GCDH, GDAP1, GFER, GFM1, GFM2, GLDC, GLRX5, GTPBP3, GYG2, HADH, HADHA, HADHB, HARS2, HCCS, HIBCH, HLCS, HMGCL, HMGCS2, HSD17B10, HSPD1, HTRA2, IARS2, IBA57, IDH2, IDH3B, IFIH1, ISCA1, ISCA2, ISCU, KARS, L2HGDH, LAMP2, LARS, LARS2, LIAS, LIPT1, LIPT2, LMBRD1, LONP1, LRPPRC, LYRM4, LYRM7, MARS2, MECR, MFF, MFN2, MGME1, MICU1, MIPEP, MPC1, MPV17, MRPL12, MRPL3, MRPL40, MRPL44, MRPS14, MRPS16, MRPS2, MRPS22, MRPS23, MRPS34, MRPS7, MSTO1, MTFMT, MTHFD1, MTO1, MTPAP, NADK2, NARS2, NAXE, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFS1, NFU1, NGLY1, NNT, NR2F1, NSUN3, NUBPL, NUP62, OGDH, OPA1, OPA3, OTC, OXCT1, PANK2, PARS2, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PET100, PINK1, PITRM1, PMPCA, PMPCB, PNKD, PNPLA8, PNPT1, POLG, POLG2, POP1, PPA2, PPOX, PSAP, PUS1, QARS, QRSL1, RANBP2, RARS, RARS2, REEP1, RMND1, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RRM2B, SACS, SAMHD1, SARS2, SCN1A, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHC, SDHD, SERAC1, SFXN4, SIRT1, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A21, SLC25A22, SLC25A26, SLC25A3, SLC25A32, SLC25A38, SLC25A4, SLC25A42, SLC25A46, SLC39A8, SLC52A2, SLC52A3, SLC6A8, SLC7A13, SPAST, SPG7, STAT2, STXBP1, SUCLA2, SUCLG1, SUCLG2, SUGCT, SURF1, TACO1, TANGO2, TARS2, TAZ, TFAM, TIMM50, TIMM8A, TIMMDC1, TK2, TMEM126A, TMEM126B, TMEM70, TOP1MT, TOP3A, TPK1, TRAP1, TREX1, TRIT1, TRMT10C, TRMT5, TRMU, TRNT1, TSFM, TTC19, TUFM, TWNK, TXN2, TYMP, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, VARS2, WARS2, WDR45, WFS1, XPNPEP3, YARS2, YME1L1
98005	代謝/ 新生児	Invitae包括的な神経代謝障害パネル Invitae Comprehensive Neurometabolic Disorders Panel	ABAT, ABCD1, ABCD4, ACAT1, ACO2, ADSL, AGA, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, AMN, AMT, AP1S1, AP4M1, APTX, ARG1, ARHGEF9, ARSA, ASAH1, ASL, ASNS, ASPA, ASS1, ATAD1, ATP13A2, ATP6AP1, ATP6AP2, ATP7A, ATP7B, AUH, BCKDHA, BCKDHB, BCKDK, BSCL2, BSND, BTD, C19ORF12, CA5A, CAD, CASR, CBS, CD320, CLCNKB, CLDN16, CLDN19, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLPB, CNNM2, COASY, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, CP, CPOX, CPS1, CRAT, CTSD, CUBN, CYP27A1, D2HGDH, DBH, DBT, DCAF17, DDC, DHCR7, DHFR, DLAT, DLD, DNAJC12, EGF, ETFA, ETFB, ETFDH, ETHE1, FA2H, FAM111A, FBXL4, FH, FOLR1, FTL, FUCA1, FXYD2, GAD1, GALC, GAMT, GATM, GCDH, GCH1, GCLC, GIF, GJA1, GLA, GLB1, GLDC, GLRA1, GLRB, GLUD1, GNS, GOT2, GPHN, GSS, GTPBP2, GUSB, HCFC1, HEXA, HEXB, HGSNAT, HLCS, HMBS, HMGCL, HMGCS2, HNF1B, HPRT1, HSD17B10, IDH2, IDS, IDUA, IVD, KCNA1, KCNJ10, KCTD7, KIF1A, L2HGDH, LIPA, LMBRD1, MAN2B1, MAOA, MAT1A, MCCC1, MCCC2, MCEE, MFSD8, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCOS, MOCS1, MOCS2, MPV17, MSMO1, MTHFR, MTR, MTRR, MUT, NAGLU, NAGS, NAXE, NGLY1, NPC1, NPC2, NT5C3A, OAT, OTC, OXCT1, PAH, PANK2, PC, PCBD1, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PGM3, PHGDH, PHYH, PLA2G6, PNP, PNPO, POLG, PPM1K, PPT1, PRDX1, PROSC, PRPS1, PSAT1, PSPH, PTS, QDPR, RAPSN, REPS1, SCN4A, SCP2, SGSH, SLC12A1, SLC12A3, SLC13A5, SLC18A2, SLC19A1, SLC19A2, SLC19A3, SLC1A3, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A42, SLC2A1, SLC30A10, SLC33A1, SLC39A14, SLC39A8, SLC46A1, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SLC6A9, SPR, SQSTM1, SUCLA2, SUCLG1, SUOX, TAT, TCN1, TCN2, TH, TPI1, TPK1, TRPM6, TTPA, TWNK, WDR45, XDH
4112	眼科	Invitaeバルデー・ビードル症候群パネル Invitae Bardet-Biedl Syndrome Panel	ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP164, CEP19, CEP290, FBN3, IFT172, IFT27, IFT74, KIF7, LZTFL1, MKKS, MKS1, SCLT1, SDCCAG8, TRAPPC3, TRIM32, TTC8, WDPCP
5132	眼科	Invitae白内障パネル Invitae Cataracts Panel	ABCA3, ABCB6, ABHD12, ADAMTS18, ADAMTSL4, AGK, ALDH18A1, BCOR, BEST1, BFSP1, BFSP2, CHMP4B, CLN3, COL11A1, COL18A1, COL2A1, COL4A1, COL4A2, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, CYP27A1, CYP51A1, EPG5, EPHA2, ERCC2, ERCC5, ERCC6, ERCC8, EYA1, FAM126A, FOXC1, FOXE3, FTL, FYCO1, FZD4, GALK1, GALT, GCNT2, GFER, GJA1, GJA3, GJA8, HMX1, HSF4, JAM3, LEMD2, LIM2, LONP1, LSS, MAF, MIP, MIR184, MYH9, NDP, NF2, NHS, OCRL, OPA3, P3H2, PAX6, PEX10, PEX11B, PEX16, PEX2, PEX7, PITX2, PITX3, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RDH11, RECQL4, RGS6, RNLS, RRAGA, SC5D, SIL1, SIPA1L3, SIX6, SLC16A12, SLC33A1, TBC1D20, TDRD7, TFAP2A, TMEM70, UNC45B, VIM, VSX2, WDR87, WFS1, WRN, XYLT2

4301	眼科	Invitae先天性停止性夜盲パネル Invitae Congenital Stationary Night Blindness Panel	CABP4, CACNA1F, CACNA2D4, CHM, CYP4V2, FRMD7, GNAT1, GNB3, GPR179, GRM6, GUCY2D, LRIT3, NYX, PDE6B, RBP4, RDH5, RHO, RLBP1, RPE65, SAG, SLC24A1, TRPM1
4302	眼科	Invitae角膜ジストロフィパネル Invitae Corneal Dystrophies Panel	CHRDL1, CHST6, COL17A1, COL5A1, COL8A2, CYP4V2, DCN, FOXE3, GJA8, GRHL2, GSN, KERA, KRT12, KRT3, LCAT, LOXHD1, MAF, MIR184, NLRP1, OVOL2, PAX6, PIKFYVE, PITX2, PRDM5, PXDN, SLC4A11, TACSTD2, TGFBI, UBIAD1, VSX1, ZEB1, ZNF143, ZNF469
55015	眼科	Invitae緑内障パネル Invitae Glaucoma Panel	ASB10, ATOH7, BMP4, COL4A1, COL8A2, CYP1B1, EXO5, FOXC1, FOXE3, LMX1B, LTBP2, MAF, MFRP, MYOC, OPTN, PAX6, PIK3R1, PITX2, PITX3, PRPF8, PRSS56, PXDN, SH3PXD2B, SIX6, SLC4A4, TEK, WDR36
72100	眼科	Invitae遺伝性網膜疾患パネル Invitae Inherited Retinal Disorders Panel	ABCA4, ABCC6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADAMTSL4, ADGRA3, ADGRV1, ADIPOR1, AGBL5, AHI1, AHR, AIPL1, ALMS1, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARSG, ASRGL1, ATF6, ATOH7, B9D1, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C10ORF11, C12ORF65, C1QTNF5, C8ORF37, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CCT2, CDH23, CDH3, CDHR1, CEP164, CEP19, CEP250, CEP290, CEP41, CEP78, CEP83, CERKL, CFAP410, CHM, CIB2, CISD2, CLCC1, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLRN1, CLUAP1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, CPLANE1, CRB1, CRX, CSPP1, CTNNA1, CTSD, CWC27, CYP4V2, DHDDS, DHX32, DHX38, DNAJC17, DRAM2, DSCAML1, DTHD1, EFEMP1, ELOVL4, EMC1, ERCC6, EXOSC2, EYS, FAM161A, FBLN5, FLVCR1, FRMD7, FSCN2, FZD4, GDF6, GNAT1, GNAT2, GNB3, GNPTG, GNS, GPR143, GPR179, GPR45, GRM6, GRN, GUCA1A, GUCA1B, GUCY2D, HARS, HGSNAT, HK1, HMCN1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT43, IFT74, IFT80, IFT81, IFT88, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, ITM2B, JAG1, KCNJ13, KCNV2, KIAA0586, KIAA1549, KIF11, KIF7, KIZ, KLHL7, LCA5, LRAT, LRIT3, LRP2, LRP5, LYST, LZTFL1, MAK, MAPKAPK3, MERTK, MFN2, MFRP, MFSD8, MIR204, MKKS, MKS1, MPDZ, MTPAP, MTTP, MYO7A, NAGLU, NBAS, NDP, NEK2, NEUROD1, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OCA2, OFD1, OPA1, OPA3, OPN1SW, OR2W3, OTX2, P3H2, PAX2, PAX6, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PITPNM3, PLA2G5, PLK4, PNPLA6, POC1B, POC5, POMGNT1, PPT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RAB28, RAX2, RBP1, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGR (ORF15), RPGRIP1, RPGRIP1L, RS1, RTN4IP1, SAG, SAMD11, SCLT1, SDCCAG8, SEMA4A, SGSH, SIX6, SLC24A1, SLC24A5, SLC45A2, SLC7A14, SNRNP200, SPATA7, SPP2, TCTN1, TCTN2, TCTN3, TEAD1, TIMM8A, TIMP3, TMED7, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TRAF3IP1, TREX1, TRIM32, TRNT1, TRPM1, TSPAN12, TTC21B, TTC8, TTLL5, TTPA, TUB, TUBGCP4, TUBGCP6, TULP1, TYR, TYRP1, UNC119, USH1C, USH1G, USH2A, VCAN, VPS13B, WDPCP, WDR19, WDR34, WFS1, WHRN, ZNF408, ZNF423, ZNF513
434345	眼科	Invitae黄斑ジストロフィーパネル Invitae Macular Dystrophy Panel	ABCA4, BEST1, C1QTNF5, CDH3, CERKL, CFI, CHST6, CNGB3, CRB1, CRX, CTNNA1, DRAM2, EFEMP1, ELOVL4, FSCN2, GUCA1B, HMCN1, IMPG1, IMPG2, KCNV2, MFSD8, NMNAT1, PRDM13, PROM1, PRPH2, RAX2, RBP3, RBP4, RDH12, RDH5, RLBP1, RP1L1, RPGRIP1, RS1, SIX6, TIMP3
5142	眼科	Invitae小眼球症、無眼球症、コロボーマ（MAC）および前眼部発育不全パネル Invitae Microphthalmia, Anophthalmia, Coloboma (MAC) and Anterior Segment Dysgenesis Panel	ABCB6, ADAMTS18, ALDH1A3, ALX1, ASPH, BCOR, BMP4, BMP7, C12ORF57, CDON, CHD7, COL4A1, CPAMD8, CRYAA, CRYBA4, CYP1B1, DCDC1, ELP4, ERCC2, ERCC5, ERCC6, FAT1, FOXC1, FOXE3, FOXL2, FRAS1, FREM1, FREM2, FZD5, GDF6, GJA1, GRIP1, HCCS, HESX1, HMGB3, HMX1, ITPR1, KERA, MAB21L2, MFRP, MIR204, MITF, NAA10, NDP, OCRL, OTX2, PAX2, PAX6, PITX2, PITX3, PORCN, PRDM5, PRSS56, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, RBP4, RERE, SALL2, SHH, SIX3, SIX6, SLC38A8, SMCHD1, SMOC1, SOX2, STRA6, TBC1D20, TENM3, TFAP2A, UBE3B, VAX1, VSX1, VSX2, WNT2B, YAP1, ZDBF2, ZIC2
4213	眼科	Invitae OFCD症候群検査 InvitaeOculo-Facio-Cardio-Dental Syndrome Test	BCOR
434348	眼科	Invitae眼皮膚白皮症パネル Invitae Oculocutaneous Albinism Panel	AP3B1, AP3D1, BLOC1S3, BLOC1S6, C10ORF11, DTNBP1, FRMD7, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, MLPH, MYO5A, OCA2, RAB27A, SLC24A5, SLC38A8, SLC45A2, TYR, TYRP1
434349	眼科	Invitae中隔視神経形成異常パネル Invitae Septo-optic Dysplasia Panel	GLI2, HESX1, OTX2, PAX6, PROP1, SOX2, SOX3, TAX1BP3

55013	眼科	InvitaeStickler症候群パネル Invitae Stickler Syndrome Panel	COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, LOXL3, LRP2, VCAN
4112	腎臓学	InvitaeBardet-Biedl症候群パネル Invitae Bardet-Biedl Syndrome Panel	ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP164, CEP19, CEP290, FBN3, IFT172, IFT27, IFT74, KIF7, LZTFL1, MKKS, MKS1, SCLT1, SDCCAG8, TRAPPC3, TRIM32, TTC8, WDPCP
4111	腎臓学	Invitae Joubert and Meckel-Gruber Syndromes Panel	AHI1, ARL13B, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, INPP5E, KIAA0586, KIF7, MKS1, MRE11, NPHP1, NPHP3, OFD1, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423
4113	腎臓学	IInvitaeネフロン癆パネル Invitae Nephronophthisis Panel	AHI1, ANKS6, CC2D2A, CEP164, CEP290, CEP83, DCDC2, GLIS2, IFT172, INVS, IQCB1, NEK8, NPHP1, NPHP3, NPHP4, OFD1, PKHD1, RPGRIP1L, SDCCAG8, TCTN1, TMEM216, TMEM237, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423
4102	腎臓学	Invitae繊毛病パネル Invitae Ciliopathies Panel	ADAMTS9, AHI1, AK7, ALG8, ALMS1, ANKS6, ARL13B, ARL3, ARL6, ARMC4, ARMC9, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C11ORF70, C2CD3, C8ORF37, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CCNQ, CELSR2, CENPF, CEP104, CEP120, CEP164, CEP19, CEP290, CEP41, CEP55, CEP83, CFAP298, CFAP410, CFTR, CLUAP1, CPLANE1, CRB2, CSPP1, CTU2, DCDC2, DDX59, DHCR7, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB11, DNAJB13, DNAL1, DRC1, DYNC2H1, DYNC2LI1, DZIP1L, EVC, EVC2, EXOC3L2, EXOC8, FAM186B, FBN3, FGFR1, FGFR2, FGFR3, GANAB, GAS8, GLIS2, HNF1B, HYLS1, ICK, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT57, IFT74, IFT80, IFT81, INPP5E, INTU, INVS, IQCB1, KIAA0556, KIAA0586, KIAA0753, KIF14, KIF7, LRP5, LRRC56, LRRC6, LRRCC1, LZTFL1, MAPKBP1, MCIDAS, MKKS, MKS1, MRE11, NEK1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PDE6D, PIAS1, PIBF1, PIH1D3, PKD2, PKHD1, PMM2, PRKCSH, RBM48, RCOR1, RPGR, RPGRIP1L, RSPH1, RSPH3, RSPH4A, RSPH9, SCLT1, SDCCAG8, SEC63, SLC30A7, SPAG1, SUFU, TBC1D32, TCTEX1D2, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TRAF3IP1, TRAPPC3, TRIM32, TTC21B, TTC26, TTC8, TXNDC15, USP9X, WDPCP, WDR19, WDR34, WDR35, WDR60, XPNPEP3, ZMYND10, ZNF423
4103	腎臓学	Invitae周産期致死骨格異形成および骨格繊毛病パネル Invitae Perinatal Lethal Skeletal Dysplasia and Skeletal Ciliopathies Panel	AGPS, ALPL, ASCC1, CEP120, CFAP410, CHUK, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, CREB3L1, CRTAP, CSPP1, DLL1, DLL3, DMRT2, DYNC2H1, DYNC2LI1, EBP, EVC, EVC2, FAM111A, FGFR2, FGFR3, GNPAT, GPX4, HES7, HSPG2, ICK, IFT122, IFT140, IFT172, IFT43, IFT52, IFT74, IFT80, IFT81, IMPAD1, INPPL1, INTU, KIAA0586, KIAA0753, LBR, LFNG, LRP5, MBTPS2, MESP2, NEK1, P3H1, PAM16, PEX5, PEX7, PPIB, RIPPLY2, SLC26A2, SLC35D1, SOX9, TBX6, TCTEX1D2, TMEM38B, TRAF3IP1, TRIP11, TRIP4, TRPV4, TTC21B, WDR19, WDR34, WDR35, WDR60, WNT1, WNT3
4101	腎臓学	Invitae原発性線毛機能不全パネル Invitae Primary Ciliary Dyskinesia Panel	AK7, ARMC4, C11ORF70, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CEP164, CFAP298, CFTR, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, GAS8, LRRC56, LRRC6, MCIDAS, NOTCH2, OFD1, PIH1D3, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, ZMYND10
434341	腎臓学	Invitae先天性腎尿路異常（CAKUT）パネル Invitae Congenital Anomalies of Kidney and Urinary Tract (CAKUT) Panel	ACE, AGT, AGTR1, BICC1, BNC2, CRKL, DCHS1, DSTYK, EYA1, FAT4, FGF20, FOXC1, FRAS1, FREM1, FREM2, GATA3, GLI3, GREB1L, HNF1B, HOXA13, HPSE2, ITGA8, LRP4, NRIP1, PAX2, PBX1, REN, RET, ROBO2, SALL1, SIX1, SIX2, SIX5, SLIT2, SOX17, TBX18, UMOD, VIPAS39, VPS33B, WNT4, WT1
55008	腎臓学	Invitae嚢胞性腎疾患パネル Invitae Cystic Kidney Disease Panel	ALG8, ANKS6, BICC1, CEP164, CEP290, CEP83, CEP89, COL4A1, CRB2, DCDC2, DICER1, DNAJB11, DZIP1L, GANAB, GLIS2, HNF1B, IFT172, INVS, IQCB1, JAG1, LRP5, MAPKBP1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD2, PKHD1, PRKCSH, RPGRIP1L, SDCCAG8, SEC61A1, SEC63, TMEM67, TSC1, TSC2, TTC21B, UMOD, VHL, WDR19, ZNF423

434347	腎臓学	Invitaeネフローゼ症候群および巣状分節性糸球体硬化症(FSGS)パネル Invitae Nephrotic Syndrome and Focal Segmental Glomerulosclerosis (FSGS) Panel	ACTN4, AMN, ANLN, APOL1, ARHGAP24, ARHGDIA, CD2AP, CFH, CLCN5, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CUBN, DGKE, DHTKD1, EMP2, FN1, INF2, ITGA3, ITGB4, KANK1, KANK2, KANK4, LAGE3, LAMA5, LAMB2, LMX1B, MAGI2, MYO1E, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, NXF5, OCRL, OSGEP, PAX2, PDSS2, PLCE1, PODXL, PTPRO, SGPL1, SMARCAL1, TP53RK, TPRKB, TRPC6, WDR73, WT1, XPO5
4308	腎臓学	Invitae腎尿細管性障害パネル Invitae Renal Tubular Disorders Panel	ACE, AGT, AGTR1, AQP2, ATP6V0A4, ATP6V1B1, AVPR2, BSND, CA2, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, CUL3, EGF, FOXI1, FXYD2, GATM, GNA11, HSD11B2, KCNJ1, KCNJ10, KLHL3, MAGED2, NR3C2, OCRL, REN, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A3, SLC34A1, SLC4A1, SLC4A4, TRPM6, WNK1, WNK4
72037	腎臓学	Invitae腎結石パネル Invitae Nephrolithiasis Panel	ADCY10, AGXT, ALPL, APRT, ATP6V0A4, ATP6V1B1, ATP7B, CA2, CASR, CLCN5, CLCNKB, CLDN16, CLDN19, CYP24A1, FAM20A, FOXI1, GPHN, GRHPR, HOGA1, HPRT1, KCNJ1, MOCOS, MOCS1, MOCS2, OCRL, PEX6, PREPL, SLC12A1, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC7A9, SLC9A3R1, UMOD, VDR, XDH
55005	腎臓学	Invitaeアルポート症候群パネル Invitae Alport Syndrome Panel	CD151, COL4A3, COL4A4, COL4A5, COL4A6, MYH9
55012	腎臓学	Invitae新生児呼吸窮迫パネル Invitae Neonatal Respiratory Distress Panel	ABCA3, ACE, AFF4, AGT, AGTR1, AK7, ALB, ARL6, ARMC4, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C11ORF70, C8ORF37, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CD40, CD40LG, CEP164, CEP19, CEP290, CFAP298, CFTR, COPA, CSF2RA, CSF2RB, CXCR4, DKC1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, ELANE, FBN3, FLNA, FOXF1, GAS8, GATA2, HSD11B2, IFT172, IFT27, IFT74, IL1RN, INPPL1, ITGA3, KIF7, LRRC56, LRRC6, LZTFL1, MARS, MCIDAS, MKKS, MKS1, MTHFR, MTM1, NDST1, NKX2-1, NME8, NOTCH2, OFD1, PARN, PIEZO2, PIH1D3, REN, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, RTEL1, SARS2, SCLT1, SDCCAG8, SFTPB, SFTPC, SLC27A4, SLC34A2, SLC7A7, SPAG1, TERC, TERT, TINF2, TMEM165, TMEM173, TRAPPC3, TRIM32, TTC8, WDPCP, ZMYND10
55682	腎臓学	Invitae非典型溶血性尿毒症症候群および血栓性微小血管症パネル Invitae Atypical Hemolytic Uremic Syndrome and Thrombotic Microangiopathies Panel	ADAMTS13, C3, CD46, CD55, CD59, CFB, CFH, CFI, DGKE, INF2, MMACHC, PLG, THBD
75000	腎臓学	Invitae進行性腎疾患パネル Invitae Progressive Renal Disease Panel	ACE, ACTB, ACTN4, ADAMTS13, AGT, AGTR1, AHI1, ALG8, ALG9, ALMS1, AMN, ANKS6, ANLN, APOL1, APRT, AQP2, ARHGAP24, ARHGDIA, ARL6, ATP6V0A4, ATP6V1B1, AVP, AVPR2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BICC1, BSND, CA2, CACNA1D, CACNA1H, CASR, CD151, CD2AP, CDKN1C, CEP164, CEP19, CEP290, CEP83, CEP89, CFHR5, CHRM3, CLCN5, CLCNKB, CLDN16, CLDN19, COL4A1, COL4A3, COL4A4, COL4A5, COL4A6, COQ2, COQ6, COQ8B, CPLANE1, CRB2, CTNS, CUBN, CYP24A1, DCDC2, DGKE, DHCR7, DICER1, DLC1, DNAJB11, DZIP1L, EGF, EMP2, EYA1, FAM20A, FAN1, FAT1, FN1, FOXI1, FOXP3, FREM1, GANAB, GATA3, GATM, GLA, GLIS2, GRHPR, GSN, HNF1A, HNF1B, HPRT1, HPSE2, HSD11B2, IFT122, IFT140, IFT172, IFT27, IFT43, INF2, INVS, IQCB1, ITGA3, ITGA6, ITGB4, JAG1, KANK2, KANK4, KAT6B, KCNJ1, KCNJ10, LAMA5, LAMB2, LCAT, LDHA, LMX1B, LPIN1, LRP4, LRP5, LYZ, LZTFL1, MAGED2, MAGI2, MAPKBP1, MEFV, MKKS, MYH9, MYO1E, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NSDHL, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, OFD1, PAX2, PBX1, PDSS2, PHEX, PKD2, PKHD1, PLCE1, PODXL, PREPL, PTPRO, REN, RMND1, RPGRIP1L, SALL1, SALL4, SARS2, SDCCAG8, SEC61A1, SEMA3E, SGPL1, SIX1, SIX5, SLC12A1, SLC12A3, SLC22A12, SLC2A2, SLC2A9, SLC37A4, SLC41A1, SLC4A1, SLC4A4, SLC7A7, SMARCAL1, SYNPO, TBC1D8B, TMEM67, TNS2, TP63, TRIM32, TRPC6, TSC1, TSC2, TTC21B, TTC8, UMOD, WDPCP, WDR19, WDR73, WT1, XPNPEP3, XPO5, ZNF423

3201	神経疾患	Invitaeシャルコー・マリー・トゥース病総合パネル Invitae Charcot-Marie-Tooth Disease Comprehensive Panel	AARS, AIFM1, ATP1A1, BAG3, BSCL2, COX6A1, DHTKD1, DNAJB2, DNM2, DRP2, DYNC1H1, EGR2, FBLN5, FGD4, FIG4, GARS, GDAP1, GJB1, GNB4, HARS, HINT1, HSPB1, HSPB8, IGHMBP2, INF2, KIF5A, LITAF, LMNA, LRSAM1, MARS, MCM3AP, MED25, MFN2, MME, MORC2, MPZ, MTMR2, NDRG1, NEFH, NEFL, PDK3, PLEKHG5, PMP2, PMP22, PRPS1, PRX, RAB7A, SBF1, SBF2, SH3TC2, SLC25A46, SPG11, SURF1, TFG, TRIM2, TRPV4, YARS
3230	神経疾患	Invitae遺伝性感覚・自律神経ニューロパシチーパネル Invitae Hereditary Sensory and Autonomic Neuropathy Panel	ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, PRDM12, RETREG1, SCN11A, SCN9A, SPTLC1, SPTLC2, WNK1
3461	神経疾患	Invitae家族性自律神経障害検査 Invitae Familial Dysautonomia Test	ELP1
3251	神経疾患	Invitae遺伝性痙性対麻痺総合パネル Invitae Hereditary Spastic Paraple gia Comprehensive Panel	ABCD1, ALDH18A1, ALS2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARG1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C12ORF65, CAPN1, CPT1C, CYP27A1, CYP2U1, CYP7B1, DDHD1, DDHD2, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, GBA2, GJC2, HACE1, HEXA, HSPD1, KCNA2, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, L1CAM, MAG, NIPA1, NKX6-2, NT5C2, PLP1, PNPLA6, RAB3GAP2, REEP1, REEP2, RTN2, SACS, SLC16A2, SPART, SPAST, SPG11, SPG21, SPG7, TECPR2, TFG, UCHL1, VAMP1, WASHC5, ZFYVE26
3240	神経疾患	Invitae遺伝性運動神経障害パネル Invitae Hereditary Motor Neuropathies Panel	ASAH1, ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, EXOSC9, FBXO38, GARS, HEXA, HINT1, HSPB1, HSPB8, IGHMBP2, MORC2, PLEKHG5, REEP1, SIGMAR1, SLC5A7, SMN1, SMN2, TRPV4, UBA1, VAPB, VRK1
3220	神経疾患	Invitae小径線維ニューロパチー検査 Invitae Small Fiber Neuropathy Test	SCN9A
3245	神経疾患	Invitae脊髄性筋萎縮症パネル Invitae Spinal Muscular Atrophy Panel	SMN1, SMN2
73000	神経疾患	Invitae脊髄性筋萎縮症STATパネル Invitae Spinal Muscular Atrophy STAT Panel	SMN1, SMN2
3291	神経疾患	Invitae包括的な筋ジストロフィーパネル Invitae Comprehensive Muscular Dystrophy Test	ANO5, B3GALNT2, B4GAT1, CAPN3, CAV3, CHKB, COL12A1, COL6A1, COL6A2, COL6A3, DAG1, DES, DMD, DNAJB6, DPM1, DPM2, DPM3, DYSF, EMD, FHL1, FKRP, FKTN, GAA, GMPPB, GOSR2, HNRNPDL, ISPD, ITGA7, LAMA2, LARGE1, LMNA, MYOT, PLEC, PNPLA2, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1, SGCA, SGCB, SGCD, SGCG, SMCHD1, TCAP, TK2, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN
3361	神経疾患	Invitae包括的なミオパチーパネル Invitae Comprehensive Myopathy Panel	ACTA1, ADSSL1, AMPD1, ANO5, ATP2A1, BAG3, BIN1, CACNA1S, CASQ1, CAV3, CCDC78, CFL2, CLCN1, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, CPT2, CRYAB, DES, DNAJB6, DNM2, DYSF, FHL1, FKBP14, FLNC, GNE, GYG1, GYS1, HACD1, HNRNPA2B1, ISCU, KBTBD13, KCNJ2, KLHL40, KLHL41, LAMP2, LDB3, LMNA, LMOD3, MAP3K20, MATR3, MEGF10, MICU1, MTM1, MYH2, MYH7, MYL2, MYO18B, MYOT, MYPN, NEB, ORAI1, PYROXD1, RYR1, SCN4A, SELENON, SMPX, SPEG, SQSTM1, STAC3, STIM1, TAZ, TIA1, TK2, TNNT1, TPM2, TPM3, TTN, VCP, VMA21

3280	神経疾患	Invitae包括的な神経筋障害パネル Invitae Comprehensive Neuromuscular Disorders Panel	ABHD5, ACAD9, ACADM, ACADVL, ACTA1, ADSSL1, AGK, AGL, AGRN, AHCY, ALDOA, ALG14, ALG2, AMACR, AMPD1, ANO5, ATP2A1, ATP7B, B3GALNT2, B4GAT1, BAG3, BIN1, C1QBP, CACNA1S, CAPN3, CASQ1, CAV3, CCDC78, CFL2, CHAT, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CLCN1, CNTN1, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, COQ2, COQ4, COQ7, COQ8A, COQ9, COX15, COX20, COX6B1, CPT1A, CPT2, CRYAB, CTDP1, DAG1, DDC, DES, DGUOK, DMD, DNA2, DNAJB6, DNM2, DOK7, DPAGT1, DPM1, DPM2, DPM3, DYSF, EMD, ENO3, ETFA, ETFB, ETFDH, FBXL4, FDX2, FHL1, FKBP14, FKRP, FKTN, FLAD1, FLNC, GAA, GATM, GBE1, GFER, GFPT1, GMPPB, GNE, GOSR2, GYG1, GYS1, HACD1, HADH, HADHA, HADHB, HMBS, HNRNPA2B1, HNRNPDL, ISCU, ISPD, ITGA7, KBTBD13, KCNJ2, KLHL40, KLHL41, LAMA2, LAMP2, LARGE1, LDB3, LDHA, LMNA, LMOD3, LPIN1, MAN2B1, MAP3K20, MATR3, MEGF10, MGME1, MICU1, MPV17, MTM1, MUSK, MYH2, MYH3, MYH7, MYL2, MYO18B, MYOT, MYPN, NEB, OPA1, OPA3, ORAI1, PDSS1, PDSS2, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PLEC, PNPLA2, PNPLA8, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PREPL, PUS1, PYGM, PYROXD1, RAPSN, RBCK1, RNASEH1, RRM2B, RXYLT1, RYR1, SCN4A, SDHA, SELENON, SGCA, SGCB, SGCD, SGCG, SIL1, SLC16A1, SLC18A3, SLC22A5, SLC25A20, SLC25A3, SLC25A4, SLC25A42, SLC5A7, SMCHD1, SMN1, SMN2, SMPX, SPEG, SQSTM1, STAC3, STIM1, SUCLA2, SUCLG1, SYT2, TANGO2, TAZ, TCAP, TIA1, TK2, TNNT1, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TRMT5, TSFM, TTN, TWNK, TYMP, VAMP1, VCP, VMA21, YARS2
3292	神経疾患	Invitae先天性筋ジストロフィーパネル Invitae Congenital Muscular Dystrophy Panel	B3GALNT2, B4GAT1, CHKB, COL12A1, COL6A1, COL6A2, COL6A3, DAG1, DMD, DPM1, DPM2, DPM3, FKRP, FKTN, GMPPB, GOSR2, ISPD, ITGA7, LAMA2, LARGE1, LMNA, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1, TCAP, TK2
3362	神経疾患	Invitae先天性ミオパチーパネル Invitae Congenital Myopathy Panel	ACTA1, BIN1, CACNA1S, CCDC78, CFL2, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, DNM2, FKBP14, HACD1, KBTBD13, KLHL40, KLHL41, LMOD3, MAP3K20, MEGF10, MICU1, MTM1, MYH7, MYL2, MYO18B, MYPN, NEB, PYROXD1, RYR1, SELENON, SPEG, STAC3, TK2, TNNT1, TPM2, TPM3, TTN
3301	神経疾患	Invitaeジスロトフィン異常症検査 Invitae Dystrophinopathies Test	DMD
53699	神経疾患	Invitae横紋筋融解症および代謝性ミオパチーパネル Invitae Rhabdomyolysis and Metabolic Myopathy Panel	ABHD5, ACAD9, ACADM, ACADVL, AGK, AGL, AHCY, ALDOA, AMACR, AMPD1, ANO5, ATP2A1, ATP7B, B3GALNT2, B4GAT1, C1QBP, CACNA1S, CAPN3, CASQ1, CAV3, CHAT, CHKB, COQ2, COQ4, COQ7, COQ8A, COQ9, COX15, COX20, COX6B1, CPT1A, CPT2, CTDP1, DAG1, DGUOK, DMD, DNA2, DNAJB6, DPM1, DPM2, DPM3, DYSF, EMD, ENO3, ETFA, ETFB, ETFDH, FBXL4, FDX2, FHL1, FKRP, FKTN, FLAD1, GAA, GATM, GBE1, GFER, GMPPB, GYG1, GYS1, HADH, HADHA, HADHB, HMBS, ISCU, ISPD, ITGA7, LAMA2, LAMP2, LARGE1, LDHA, LPIN1, MAN2B1, MGME1, MICU1, MPV17, MYH3, OPA1, OPA3, PDSS1, PDSS2, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PNPLA2, PNPLA8, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PUS1, PYGM, RBCK1, RNASEH1, RRM2B, RXYLT1, RYR1, SCN4A, SDHA, SGCA, SGCB, SGCD, SGCG, SIL1, SLC16A1, SLC22A5, SLC25A20, SLC25A3, SLC25A4, SLC25A42, STAC3, SUCLA2, SUCLG1, TANGO2, TCAP, TK2, TNPO3, TRIM32, TRMT5, TSFM, TWNK, TYMP, YARS2
3304	神経疾患	Invitae肢帯型筋ジストロフィーパネル Invitae Limb-Girdle Muscular Dystrophy Panel	ANO5, CAPN3, CAV3, DAG1, DES, DMD, DNAJB6, DYSF, FKRP, FKTN, GAA, GMPPB, GOSR2, HNRNPDL, ISPD, LAMA2, LMNA, MYOT, PLEC, PNPLA2, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, SGCA, SGCB, SGCD, SGCG, SMCHD1, TCAP, TK2, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN
3285	神経疾患	Invitae悪性高熱症感受性パネル Invitae Malignant Hyperthermia Susceptibility Panel	CACNA1S, RYR1, STAC3
3375	神経疾患	Invitae筋強直および先天性パラミオトニアパネル Invitae Myotonia and Paramyotonia Congenita Panel	CLCN1, SCN4A
3373	神経疾患	Invitae周期性四肢麻痺パネル Invitae Periodic Paralysis Panel	ATP1A2, CACNA1S, KCNJ2, MCM3AP, RYR1, SCN4A

3351	神経疾患	Invitaeジストニア総合パネル Invitae Dystonia Comprehensive Panel	ACTB, ADCY5, ANO3, ATP1A3, ATP7B, BCAP31, CIZ1, COL6A3, CYP27A1, GCH1, GNAL, GNAO1, HEXA, HPCA, KCNMA1, KCTD17, KMT2B, MECR, PANK2, PLA2G6, PNKD, PRKN, PRKRA, PRRT2, SGCE, SLC2A1, SLC30A10, SLC39A14, SLC6A3, SPR, TH, THAP1, TOR1A, TUBB4A, VAC14, VPS13A, VPS13D, XPR1
3503	神経疾患	Invitae筋萎縮性側索硬化症パネル Invitae Amyotrophic Lateral Sclerosis Panel	ALS2, ANG, ANXA11, CHCHD10, DCTN1, ERBB4, FUS, HEXA, KIF5A, OPTN, PFN1, SETX, SOD1, SPG11, SQSTM1, TARDBP, TBK1, TFG, UBQLN2, VAPB, VCP
3505	神経疾患	Invitae前頭側頭型認知症パネル Invitae Frontotemporal Dementia Panel	CHCHD10, CHMP2B, DCTN1, FUS, GRN, HNRNPA2B1, MAPT, SQSTM1, TARDBP, TBK1, TREM2, UBQLN2, VCP
3504	神経疾患	Invitae遺伝性アルツハイマー病パネル Invitae Hereditary Alzheimer's Disease Panel	APP, PSEN1, PSEN2
3502	神経疾患	Invitae遺伝性筋萎縮性側索硬化症、前頭側頭型認知症およびアルツハイマー病パネル Invitae Hereditary Amyotrophic Lateral Sclerosis, Frontotemporal Dementia and Alzheimer Disease Panel	ALS2, ANG, ANXA11, APP, CHCHD10, CHMP2B, DCTN1, ERBB4, FUS, GRN, HEXA, HNRNPA2B1, ITM2B, KIF5A, MAPT, OPTN, PFN1, PRNP, PSEN1, PSEN2, SETX, SNCA, SOD1, SORL1, SPG11, SQSTM1, TARDBP, TBK1, TFG, TREM2, UBQLN2, VAPB, VCP
3352	神経疾患	Invitae遺伝性パーキンソン病およびパーキンソニズムパネル Invitae Hereditary Parkinson Disease and Parkinsonism Panel	ATP13A2, ATP7B, CHCHD2, CSF1R, DCTN1, DNAJC6, FBXO7, GBA, GCH1, LRRK2, PARK7, PDE8B, PINK1, PLA2G6, PRKN, PRKRA, RAB39B, SLC6A3, SNCA, SPR, SYNJ1, TH, TMEM230, VPS13C, VPS35, XPR1
3506	神経疾患	Invitaeプリオン病検査 Invitae Hereditary Prion Disease Test	PRNP
53700	神経疾患	Invitae家族性片麻痺性片頭痛パネル Invitae Familial Hemiplegic Migraine Panel	ATP1A2, ATP1A3, CACNA1A, PRRT2, SCN1A, SLC1A3, SLC2A1
53701	神経疾患	Invitae遺伝性脳小血管疾患パネル Invitae Hereditary Cerebral Small Vessel Disease Panel	APP, CBS, COL4A1, COL4A2, CST3, FOXC1, GLA, HTRA1, NOTCH3, TREX1
53702	神経疾患	Invitae遺伝性もやもや病パネル Invitae Hereditary Moyamoya Disease Panel	GUCY1A1, RNF213
4741	神経疾患	Invitae Baraitser-Winter脳前頭顔症候群パネル Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Test	ACTB, ACTG1
55006	神経疾患	Invitae脳奇形パネル Invitae Brain Malformations Panel	ACTB, ACTG1, ADGRG1, ADNP, AHDC1, AKT3, AMPD2, APC2, ARFGEF2, ARID1A, ARID1B, ARX, ASNS, ASPM, ATP6V0A2, B3GALNT2, B4GAT1, BMP4, C19ORF12, CASK, CCM2, CCND2, CDK13, CDK5, CDON, CHMP1A, CIT, CNOT1, CNOT3, COASY, COL18A1, COL3A1, COL4A1, COL4A2, CP, CRADD, CUL4B, DAG1, DCHS1, DCX, DIAPH1, DISP1, DLL1, DMXL2, DPF2, DYNC1H1, EIF2AK2, EMC1, EML1, ERMARD, EXOSC3, FA2H, FAT4, FGFR1, FIG4, FKRP, FKTN, FLNA, FOXA2, FTL, GAS1, GLI2, GMPPB, GPSM2, GRIN2B, HIVEP2, IER3IP1, IQSEC2, ISPD, KATNB1, KCNMA1, KIF11, KIF1BP, KIF2A, KIF7, KMT2E, KRIT1, L1CAM, LAMA1, LAMB1, LAMC3, LARGE1, LRP2, MACF1, MED12, MED17, MFSD2A, MRE11, NDE1, NEDD4L, NPRL3, OCLN, OPHN1, PAFAH1B1, PANK2, PDCD10, PHGDH, PIK3R2, PLA2G6, PNKP, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPP1R12A, PTCH1, RAB11B, RAB18, RAB3GAP1, RAB3GAP2, RAD21, RARS2, RBM10, RELN, RERE, RTTN, RXYLT1, SEPSECS, SHH, SIN3A, SIX3, SLC25A19, SMARCA4, SMARCB1, SMARCE1, SMC1A, SNAP29, SON, SOX2, SRD5A3, STAG2, STAMBP, STIL, TBC1D20, TGIF1, TMTC3, TOE1, TRRAP, TSEN2, TSEN34, TSEN54, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP6, UBE3B, USP7, USP9X, VLDLR, VPS13A, VRK1, WDR45, WDR62, YWHAE, ZBTB18, ZBTB20, ZIC2, ZMIZ1

4422	神経疾患	Invitae脳海綿状血管腫パネル Invitae Cerebral Cavernous Malformations Panel	CCM2, KRIT1, PDCD10
3404	神経疾患	Invitae Rett/Angelmanおよび関連障害パネル Invitae Rett/Angelman and Related Disorders Panel	ADSL, ALDH5A1, ARX, ATRX, CAMK2B, CDKL5, CLTC, CNTNAP2, CTNNB1, DDX3X, DYRK1A, EHMT1, FOLR1, FOXG1, GABBR2, GRIA3, GRIN2A, GRIN2B, HDAC8, IQSEC2, KANSL1, KCNA2, MBD5, MECP2, MEF2C, NGLY1, NRXN1, SATB2, SCN2A, SCN8A, SLC6A1, SLC9A6, SMC1A, STXBP1, SYNGAP1, TBL1XR1, TCF4, UBE3A, WDR45, ZEB2
4424	神経疾患	Invitae全前脳症パネル Invitae Holoprosencephaly Panel	FGFR1, GLI2, SHH, SIX3, TGIF1, ZIC2
55002	神経疾患	Invitae白質ジストロフィーと遺伝性白質脳症パネル Invitae Leukodystrophy and Genetic Leukoencephalopathy Panel	AARS, AARS2, ABAT, ABCA1, ABCD1, ABCD4, ABHD5, ACADS, ACBD5, ACER3, ACO2, ACOX1, ACP5, ACSF3, ACTA2, ACTB, ACY1, ADAR, ADGRG1, ADK, ADNP, ADSL, AGA, AHDC1, AHI1, AIFM1, AIMP1, AIMP2, ALDH18A1, ALDH3A2, ALDH5A1, ALDH6A1, ALG12, ALG13, ALG2, ALG6, ALG9, AMACR, AMPD2, AMT, ANK3, AP1S2, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APC2, APOPT1, ARCN1, ARFGEF2, ARHGAP31, ARHGEF9, ARNT2, ARSA, ARX, ASL, ASNS, ASPA, ASS1, ASXL1, ASXL2, ATP13A2, ATP5A1, ATP6AP2, ATP6V1A, ATP7A, ATP7B, ATP8A2, ATPAF2, ATRN, ATRX, AUH, B3GALNT2, B4GALNT1, BCAP31, BCAT2, BCKDHA, BCKDHB, BCL11B, BCS1L, BICD2, BMP4, BOLA3, BPTF, BRAT1, BTD, C12ORF57, C12ORF65, C19ORF12, C2CD3, CACNA1A, CACNA1E, CARS2, CBS, CC2D2A, CCDC88A, CDC42, CDKL5, CEP290, CHMP1A, CLCN2, CLCN4, CLCN7, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLP1, CLPP, CNNM2, CNOT1, CNTNAP1, CNTNAP2, COA7, COASY, COG7, COL18A1, COL3A1, COL4A1, COL4A2, COLGALT1, COQ2, COQ6, COQ7, COQ8A, COQ9, COX10, COX14, COX15, COX20, COX6B1, COX7B, COX8A, CPLANE1, CPLX1, CPS1, CRAT, CREBBP, CRIPT, CRLF1, CSF1R, CSPP1, CTBP1, CTC1, CTDP1, CTNS, CTSA, CTSD, CUL4B, CYB5R3, CYFIP2, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS, DARS2, DBT, DCAF17, DCX, DDC, DDHD1, DDHD2, DDOST, DDX3X, DEAF1, DEGS1, DGUOK, DHCR24, DHFR, DHX37, DLD, DLL4, DMXL2, DNM1L, DNM2, DOCK6, DOCK7, DOLK, DONSON, DPAGT1, DPM1, DPM2, DPM3, DPYS, DYRK1A, EARS2, EDNRB, EHMT1, EIF2AK1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL1, ELOVL4, ENTPD1, EOGT, EPG5, EPRS, ERCC1, ERCC2, ERCC3, ERCC6, ERCC8, ETFA, ETFB, ETFDH, ETHE1, EXOSC2, EXOSC3, EXOSC8, EXOSC9, FA2H, FAM126A, FAR1, FARS2, FARSB, FASTKD2, FBXL4, FDX2, FGFRL1, FH, FIG4, FKRP, FKTN, FLVCR2, FOLR1, FOXC1, FOXG1, FOXRED1, FUCA1, FUK, GAA, GABBR2, GABRA1, GABRB1, GABRB2, GABRB3, GALC, GALT, GAN, GATAD2B, GCDH, GDAP1, GFAP, GFM1, GFM2, GJA1, GJB1, GJC2, GLA, GLB1, GLDC, GLRX5, GLUL, GLYCTK, GM2A, GMNN, GNAO1, GNS, GOT2, GPHN, GRIN1, GRM7, GTF2H5, GTPBP2, HACE1, HCN1, HEPACAM, HERC1, HEXA, HEXB, HIBCH, HIKESHI, HK1, HLCS, HMGCL, HNRNPU, HSD17B10, HSD17B4, HSPD1, IARS, IARS2, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, INPP5E, IREB2, ISCA1, ISCA2, ITPA, ITPR1, IVD, JAM3, KARS, KAT6B, KATNB1, KCNJ10, KCNJ2, KCNMA1, KCNT1, KCTD7, KDM1A, KIAA0556, KIAA0586, KIAA0753, KIDINS220, KIF1A, KIF1C, KLHL7, KMT2E, L2HGDH, LAGE3, LAMA1, LAMA2, LAMB1, LARGE1, LARS2, LETM1, LIAS, LIPT1, LIPT2, LONP1, LRPPRC, LYRM7, MAG, MAGEL2, MAN2B1, MANBA, MARS2, MAST1, MAT1A, MCCC1, MCCC2, MCOLN1, MDH2, MECP2, MED17, MED25, MEF2C, MFSD2A, MFSD8, MGP, MICU1, MKS1, MLC1, MLYCD, MMACHC, MMADHC, MOCS1, MOCS2, MOGS, MORC2, MPLKIP, MPV17, MPZ, MRPL12, MRPL44, MRPS16, MRPS22, MRPS34, MSTO1, MTFMT, MTHFR, MTHFS, MTO1, MTOR, MTR, MTRR, MUT, NACC1, NADK2, NAGA, NAGLU, NAGS, NANS, NARS2, NAXD, NAXE, NDRG1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB10, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEK1, NFE2L2, NFU1, NGLY1, NKX6-2, NOTCH1, NPC1, NPC2, NPHP1, NRXN1, NSUN3, NT5C2, NTRK2, NUBPL, NUP62, OAT, OCRL, OPA1, OPA3, OSGEP, OTC, PACS1, PAFAH1B1, PAH, PANK2, PARS2, PAX1, PC, PCCA, PCCB, PCDH12, PCGF2, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGK1, PHGDH, PHYH, PIGA, PIGB, PIGG, PIGL, PIGM, PIGN, PIGP, PIGQ, PIGT, PIGU, PIGV, PIK3C2A, PIK3R2, PLA2G6, PLAA, PLEKHG2, PLP1, PMM2, PMP22, PNKP, PNPT1, POLG, POLG2, POLR1A, POLR1C, POLR3A, POLR3B, POMGNT1, POMK, POMT1, POMT2, PPP1R15B, PPP2R1A, PPP3CA, PPT1, PRF1, PRKDC, PRODH, PROSC, PRPS1, PRUNE1, PSAP, PSAT1, PSPH, PTEN, PTPN23, PURA, PUS3, PYCR2, QARS, QRSL1, RAB11B, RAB3GAP1, RAB3GAP2, RARS, RARS2, RBPJ, REPS1, RERE, RHOBTB2, RIN2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RPGRIP1L, RPIA, RPS6KC1, RRM2B, RTTN, RXYLT1, SAMD9L, SAMHD1, SCN3A, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SEPSECS, SERAC1, SETBP1, SGSH, SH3TC2, SHOC2, SHPK, SLC12A2, SLC12A5, SLC12A6, SLC13A3, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC1A2, SLC1A3, SLC1A4, SLC25A1, SLC25A12, SLC25A15, SLC25A19, SLC25A22, SLC25A4, SLC25A42, SLC25A46, SLC2A1, SLC30A10, SLC33A1, SLC35A2, SLC39A14, SLC39A8, SLC46A1, SLC6A19, SLC6A8, SLC6A9, SLC9A1, SLC9A6, SMC1A, SNAP29, SNIP1, SNORD118, SNRPB, SNX14, SOD1, SON, SOX10, SOX2, SPART, SPAST, SPATA5, SPG11, SPG7, SPTAN1, SQSTM1, SRD5A3, SSR4, ST3GAL5, STAG2, STAMBP, STAT1, STAT2, STN1, STRADA, STX11, STXBP1, STXBP2, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYNE1, SYNJ1, TACO1, TAF2, TANGO2, TARS2, TBC1D24, TBCD, TBCE, TBCK, TBX1, TCF4, TCTN2, TIMM50, TIMMDC1, TM4SF20, TMEM106B, TMEM126B, TMEM165, TMEM216, TMEM67, TMEM70, TMTC3, TOE1, TP53RK, TPI1, TPK1, TRAPPC11, TRAPPC9, TREX1, TRMT10A, TRMT5, TSC1, TSC2, TSEN54, TTC19, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUFM, TWNK, TXN2, TYMP, UBA5, UBE2A, UBE3A, UFM1, UNC13D, UPB1, USP7, VARS2, VPS11, VPS13D, VPS33A, WARS2, WDR45, WDR73, WHSC1, WWOX, YARS, YME1L1, ZEB2, ZFYVE26, ZIC1, ZNF335

434349	神経疾患	Invitae中隔視神経形成異常パネル Invitae Septo-optic Dysplasia Panel	GLI2, HESX1, OTX2, PAX6, PROP1, SOX2, SOX3, TAX1BP3
1721	神経疾患	Invitae結節性硬化症複合パネル Invitae Tuberous Sclerosis Complex Panel	TSC1, TSC2
3200	神経疾患	包括的なニューロパシーパネル Invitae Comprehensive Neuropathies Panel	AARS, AIFM1, APOA1, ASAH1, ATL1, ATL3, ATP1A1, ATP7A, BAG3, BICD2, BSCL2, CHCHD10, COX6A1, CYP27A1, CYP7B1, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1 , EXOSC9, FBLN5, FBXO38, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GLA, GNB4, GSN, HARS, HEXA, HINT1, HMBS, HSPB1, HSPB8, IGHMBP2, INF2, KIF1A, KIF5A, LITAF, LMNA , LRSAM1, MARS, MCM3AP, MED25, MFN2, MME, MORC2, MPZ, MTMR2, NDRG1, NEFH, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP2, PMP22, POLG, POLG2, PRDM12, PRPS1, PRX, RAB7A,REEP1, RETREG1, SBF1, SBF2, SCN11A, SCN9A, SEPT9, SH3TC2, SIGMAR1, SLC12A6, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SMN1, SMN2, SPG11, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4 , TTR, UBA1, VAPB, VRK1, WNK1, YARS
3281	神経疾患	Invitae先天性筋無力症候群パネル Invitae Congenital Myasthenic Syndrome Panel	AGRN, ALG14, ALG2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, GMPPB, MUSK, PREPL, RAPSN, SLC18A3, SLC5A7, SYT2, VAMP1
3401	神経疾患	Invitaeてんかんパネル Invitae Epilepsy Panel	AARS, ABAT, ADAR, ADSL, ALDH5A1, ALDH7A1, ALG1, ALG12, ALG13, ALG6, AMACR, AMT, AP2M1, AP3B2, ARG1, ARHGEF9, ARSA, ARX, ASAH1, ASNS, ATAD1, ATP1A1, ATP1A2, ATP1A3, ATP6AP2, ATP7A, ATRX, BRAT1, C12ORF57, CACNA1A, CACNA1B, CACNA1E, CACNA2D2, CAD, CAMK2B, CARS2, CASK, CCDC88A, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN4, CLCN6, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLTC, CNTN2, CNTNAP2, COG5, COL18A1, CSTB, CTNNB1, CTSD, CYFIP2, CYP27A1, DDC, DDX3X, DEAF1, DENND5A, DEPDC5, DHDDS, DHFR, DIAPH1, DMXL2, DNAJC5, DNM1, DNM1L, DOCK7, DYNC1H1, DYRK1A, ECHS1, EEF1A2, EHMT1, EMC1, EPM2A, FAR1, FARS2, FBXO11, FGF12, FLNA, FOLR1, FOXG1, FOXP1, FRRS1L, GABBR2, GABRA1, GABRA2, GABRB1, GABRB2, GABRB3, GABRG2, GAMT, GATAD2B, GATM, GCH1, GLDC, GLRA1, GLRB, GNAO1, GNB1, GOSR2, GPAA1, GPHN, GRIA3, GRIN1, GRIN2A, GRIN2B, GRIN2D, GTPBP3, HCN1, HDAC8, HEXA, HNRNPU, IER3IP1, IFIH1, IQSEC2, ITPA, KANSL1, KCNA1, KCNA2, KCNB1, KCNC1, KCND2, KCNH1, KCNH2, KCNH5, KCNJ10, KCNK4, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCTD7, KIF1A, KIF2A, KIF5A, KPNA7, LAMC3, LGI1, LIAS, MBD5, MDH2, MECP2, MEF2C, MFSD8, MICAL1, MOCS1, MOCS2, MTOR, NACC1, NAGLU, NECAP1, NEDD4L, NEXMIF, NGLY1, NHLRC1, NPC1, NPC2, NPRL3, NRXN1, NTRK2, NUS1, PACS1, PACS2, PAFAH1B1, PCDH19, PCLO, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PHGDH, PIGA, PIGB, PIGG, PIGN, PIGO, PIGP, PIGQ, PIGV, PIGW, PLAA, PLCB1, PNKD, PNKP, PNPO, PNPT1, POLG, PPP2CA, PPP2R1A, PPP2R5D, PPP3CA, PPT1, PRICKLE1, PRIMA1, PROSC, PRRT2, PSAP, PSAT1, PSPH, PTEN, PTPN23, PURA, QARS, QDPR, RAB11A, RAB11B, RAI1, RALA, RANBP2, RELN, RFT1, RHOBTB2, RNASEH2A, RNASEH2B, RNASEH2C, RNF13, ROGDI, RORB, RUSC2, SAMHD1, SATB2, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SCP2, SERPINI1, SETBP1, SGCE, SGSH, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC1A2, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A5, SLC6A8, SLC9A6, SMC1A, SNAP25, SNX27, SPATA5, SPTAN1, ST3GAL3, ST3GAL5, STAG2, STRADA, STX1B, STXBP1, STXBP2, SUMF1, SUOX, SURF1, SYN1, SYNGAP1, SYNJ1, SZT2, TANGO2, TBC1D24, TBCK, TBL1XR1, TCF4, TH, TK2, TPK1, TREX1, TRIM8, TSC1, TSC2, TSFM, TUBB2A, UBA5, UBE3A, UNC80, WDR45, WWOX, YWHAG, ZDHHC9, ZEB2, ZSWIM6
444004	神経疾患	Invitae C9orf72を伴う遺伝性ALS、前頭側頭型認知症およびアルツハイマー病パネル Invitae Hereditary Amyotrophic Lateral Sclerosis, Frontotemporal Dementia and Alzheimer Disease with C9orf72 Panel	ALS2, ANG, ANXA11, APP, C9ORF72, CHCHD10, CHMP2B, DCTN1, ERBB4, FUS, GRN, HEXA, HNRNPA2B1, ITM2B, KIF5A, MAPT, OPTN, PFN1, PRNP, PSEN1, PSEN2, SETX, SNCA, SOD1, SORL1, SPG11, SQSTM1, TARDBP, TBK1, TFG, TREM2, UBQLN2, VAPB, VCP

728434	神経疾患	Invitae神経発達障害(NDD)パネル Invitae Neurodevelopmental Disorders (NDD) Panel	ACTB, ACTG1, ADNP, ADSL, AGA, AHDC1, ALDH5A1, ALDH7A1, AMER1, ANKRD11, AP1S2, ARG1, ARID1A, ARID1B, ARSA, ARX, ASNS, ASXL1, ATP1A3, ATP7A, ATRX, AUTS2, BCAP31, BRAF, BRAT1, BRD4, BRWD3, CACNA1A, CACNA1E, CAMK2B, CASK, CBL, CC2D2A, CDK13, CDKL5, CHD2, CHD7, CHD8, CLCN4, CLN2 (TPP1), CLN3, CLN5, CLN6, CLTC, CNTNAP2, COL4A1, CREBBP, CTNNB1, CUL3, DDC, DDX3X, DEAF1, DHCR7, DNM1L, DNMT3A, DOCK6, DPF2, DYNC1H1, DYRK1A, EEF1A2, EFTUD2, EHMT1, EP300, EZH2, FGD1, FOLR1, FOXG1, FOXP1, GABBR2, GABRB3, GABRG2, GALC, GAMT, GATAD2B, GATM, GLB1, GM2A, GNAO1, GNAS, GNS, GPC3, GRIA3, GRIN1, GRIN2A, GRIN2B, HDAC8, HEXA, HEXB, HGSNAT, HIVEP2, HNRNPK, HNRNPU, HRAS, HUWE1, IDS, IDUA, IGF1R, IL1RAPL1, IQSEC2, ITPR1, KANSL1, KAT6A, KAT6B, KCNA2, KCNB1, KCNH1, KCNQ2, KCNT1, KDM5C, KDM6A, KIF1A, KMT2A, KMT2B, KMT2D, KMT2E, KRAS, L1CAM, LZTR1, MAGEL2, MAN1B1, MAP2K1, MAP2K2, MBD5, MECP2, MED12, MED13L, MEF2C, MFSD8, MID1, MTOR, NAA10, NAA15, NAGLU, NALCN, NEXMIF, NF1, NFIA, NFIX, NGLY1, NHS, NIPBL, NONO, NPC1, NR2F1, NRAS, NRXN1, NSD1, NSUN2, OCRL, OPHN1, OTC, PACS1, PACS2, PAH, PCBD1, PCDH19, PDHA1, PGAP3, PHF21A, PHF6, PHIP, PLA2G6, PMM2, POLG, PPM1D, PPP1CB, PPP2R1A, PPP2R5D, PPP3CA, PPT1, PQBP1, PTEN, PTPN11, PTS, PURA, QDPR, RAD21, RAF1, RAI1, RBM10, RIT1, RPS6KA3, SATB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SETBP1, SETD5, SGSH, SHOC2, SIN3A, SLC13A5, SLC16A2, SLC2A1, SLC6A1, SLC6A8, SLC9A6, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SON, SOS1, SOS2, SOX11, SPAST, SPATA5, SPTAN1, STAG1, STXBP1, SURF1, SYNGAP1, TAF1, TBCK, TBL1XR1, TCF20, TCF4, TELO2, TRAPPC9, TRRAP, TSC1, TSC2, TUBA1A, UBE3A, UNC80, USP9X, VPS13B, WDR45, WWOX, ZBTB18, ZBTB20, ZC4H2, ZDHHC9, ZEB2, ZIC2, ZMIZ1, ZMYND11
1745	小児 遺伝学	Invitae慢性膵炎パネル Invitae Chronic Pancreatitis Panel	CASR, CFTR, CPA1, CTRC, PRSS1, SPINK1, TRPV6
1104	小児 遺伝学	Invitae 遺伝性小児固形腫瘍パネル Invitae Hereditary Pediatric Solid Tumors Panel	AIP, ALK, APC, BAP1, BLM, BMPR1A, BUB1B, CDC73, CDK4, CDKN1B, CDKN1C, CDKN2A, CEP57, DICER1, DIS3L2, EPCAM, EXT1, EXT2, FH, GPC3, HRAS, LZTR1, MAX, MEN1, MITF, MLH1, MSH2, MSH6, NBN, NF1, NF2, PHOX2B, PMS2, POT1, PRKAR1A, PTCH1, PTEN, RB1, RECQL4, REST, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TMEM127, TP53, TRIM28, TRIP13, TSC1, TSC2, VHL, WRN, WT1
55011	小児 遺伝学	Invitae 過成長症候群パネル Invitae Overgrowth Syndromes Panel	ABCC9, AKT2, AKT3, ASPA, ASXL2, BRWD3, CCND2, CDKN1C, CHD4, CHD8, CUL4B, DICER1, DIS3L2, DNMT3A, EED, EZH2, GFAP, GLI3, GNAS, GPC3, HEPACAM, HERC1, KPTN, L1CAM, MED12, MLC1, MPDZ, MTOR, NFIA, NFIX, NONO, NPR2, NSD1, OFD1, PDGFRB, PHF21A, PIK3R2, PPP2R5B, PPP2R5C, PPP2R5D, PTCH1, PTEN, RAB39B, RASA1, RIN2, RNF125, SETD2, STRADA, SYN1, TBC1D7, TCF20, UPF3B, ZBTB20
4501	小児 遺伝学	Invitae過成長および巨頭症症候群パネル Invitae Overgrowth and Macrocephaly Syndromes Panel	AKT2, AKT3, CDKN1C, CUL4B, DIS3L2, DNMT3A, EZH2, GLI3, GPC3, KPTN, MED12, MTOR, NF1, NFIX, NPR2, NSD1, PHF6, PIK3R2, PTEN, SETD2, SPRED1
55006	小児 遺伝学	Invitae脳奇形パネル Invitae Brain Malformations Panel	ACTB, ACTG1, ADGRG1, ADNP, AHDC1, AKT3, AMPD2, APC2, ARFGEF2, ARID1A, ARID1B, ARX, ASNS, ASPM, ATP6V0A2, B3GALNT2, B4GAT1, BMP4, C19ORF12, CASK, CCM2, CCND2, CDK13, CDK5, CDON, CHMP1A, CIT, CNOT1, CNOT3, COASY, COL18A1, COL3A1, COL4A1, COL4A2, CP, CRADD, CUL4B, DAG1, DCHS1, DCX, DIAPH1, DISP1, DLL1, DMXL2, DPF2, DYNC1H1, EIF2AK2, EMC1, EML1, ERMARD, EXOSC3, FA2H, FAT4, FGFR1, FIG4, FKRP, FKTN, FLNA, FOXA2, FTL, GAS1, GLI2, GMPPB, GPSM2, GRIN2B, HIVEP2, IER3IP1, IQSEC2, ISPD, KATNB1, KCNMA1, KIF11, KIF1BP, KIF2A, KIF7, KMT2E, KRIT1, L1CAM, LAMA1, LAMB1, LAMC3, LARGE1, LRP2, MACF1, MED12, MED17, MFSD2A, MRE11, NDE1, NEDD4L, NPRL3, OCLN, OPHN1, PAFAH1B1, PANK2, PDCD10, PHGDH, PIK3R2, PLA2G6, PNKP, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPP1R12A, PTCH1, RAB11B, RAB18, RAB3GAP1, RAB3GAP2, RAD21, RARS2, RBM10, RELN, RERE, RTTN, RXYLT1, SEPSECS, SHH, SIN3A, SIX3, SLC25A19, SMARCA4, SMARCB1, SMARCE1, SMC1A, SNAP29, SON, SOX2, SRD5A3, STAG2, STAMBP, STIL, TBC1D20, TGIF1, TMTC3, TOE1, TRRAP, TSEN2, TSEN34, TSEN54, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP6, UBE3B, USP7, USP9X, VLDLR, VPS13A, VRK1, WDR45, WDR62, YWHAE, ZBTB18, ZBTB20, ZIC2, ZMIZ1

3404	小児 遺伝学	Invitae Rett/Angelmanおよび関連障害パネル Invitae Rett/Angelman and Related Disorders Panel	ADSL, ALDH5A1, ARX, ATRX, CAMK2B, CDKL5, CLTC, CNTNAP2, CTNNB1, DDX3X, DYRK1A, EHMT1, FOLR1, FOXG1, GABBR2, GRIA3, GRIN2A, GRIN2B, HDAC8, IQSEC2, KANSL1, KCNA2, MBD5, MECP2, MEF2C, NGLY1, NRXN1, SATB2, SCN2A, SCN8A, SLC6A1, SLC9A6, SMC1A, STXBP1, SYNGAP1, TBL1XR1, TCF4, UBE3A, WDR45, ZEB2
434349	小児 遺伝学	Invitae中隔視神経形成異常パネル Invitae Septo-optic Dysplasia Panel	GLI2, HESX1, OTX2, PAX6, PROP1, SOX2, SOX3, TAX1BP3
1721	小児 遺伝学	Invitae結節性硬化症複合パネル Invitae Tuberous Sclerosis Complex Panel	TSC1, TSC2
4112	小児 遺伝学	InvitaeBardet-Biedl症候群パネル Invitae Bardet-Biedl Syndrome Panel	ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP164, CEP19, CEP290, FBN3, IFT172, IFT27, IFT74, KIF7, LZTFL1, MKKS, MKS1, SCLT1, SDCCAG8, TRAPPC3, TRIM32, TTC8, WDPCP
4102	小児 遺伝学	Invitae繊毛病パネル Invitae Ciliopathies Panel	ADAMTS9, AHI1, AK7, ALG8, ALMS1, ANKS6, ARL13B, ARL3, ARL6, ARMC4, ARMC9, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C11ORF70, C2CD3, C8ORF37, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CCNQ, CELSR2, CENPF, CEP104, CEP120, CEP164, CEP19, CEP290, CEP41, CEP55, CEP83, CFAP298, CFAP410, CFTR, CLUAP1, CPLANE1, CRB2, CSPP1, CTU2, DCDC2, DDX59, DHCR7, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB11, DNAJB13, DNAL1, DRC1, DYNC2H1, DYNC2LI1, DZIP1L, EVC, EVC2, EXOC3L2, EXOC8, FAM186B, FBN3, FGFR1, FGFR2, FGFR3, GANAB, GAS8, GLIS2, HNF1B, HYLS1, ICK, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT57, IFT74, IFT80, IFT81, INPP5E, INTU, INVS, IQCB1, KIAA0556, KIAA0586, KIAA0753, KIF14, KIF7, LRP5, LRRC56, LRRC6, LRRCC1, LZTFL1, MAPKBP1, MCIDAS, MKKS, MKS1, MRE11, NEK1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PDE6D, PIAS1, PIBF1, PIH1D3, PKD2, PKHD1, PMM2, PRKCSH, RBM48, RCOR1, RPGR, RPGRIP1L, RSPH1, RSPH3, RSPH4A, RSPH9, SCLT1, SDCCAG8, SEC63, SLC30A7, SPAG1, SUFU, TBC1D32, TCTEX1D2, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TRAF3IP1, TRAPPC3, TRIM32, TTC21B, TTC26, TTC8, TXNDC15, USP9X, WDPCP, WDR19, WDR34, WDR35, WDR60, XPNPEP3, ZMYND10, ZNF423
4113	小児 遺伝学	InvitaeNephronophthisisパネル Invitae Nephronophthisis Panel	AHI1, ANKS6, CC2D2A, CEP164, CEP290, CEP83, DCDC2, GLIS2, IFT172, INVS, IQCB1, NEK8, NPHP1, NPHP3, NPHP4, OFD1, PKHD1, RPGRIP1L, SDCCAG8, TCTN1, TMEM216, TMEM237, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423
4103	小児 遺伝学	Invitae周産期致死骨格異形成および骨格繊毛病パネル Invitae Perinatal Lethal Skeletal Dysplasia and Skeletal Ciliopathies Panel	AGPS, ALPL, ASCC1, CEP120, CFAP410, CHUK, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, CREB3L1, CRTAP, CSPP1, DLL1, DLL3, DMRT2, DYNC2H1, DYNC2LI1, EBP, EVC, EVC2, FAM111A, FGFR2, FGFR3, GNPAT, GPX4, HES7, HSPG2, ICK, IFT122, IFT140, IFT172, IFT43, IFT52, IFT74, IFT80, IFT81, IMPAD1, INPPL1, INTU, KIAA0586, KIAA0753, LBR, LFNG, LRP5, MBTPS2, MESP2, NEK1, P3H1, PAM16, PEX5, PEX7, PPIB, RIPPLY2, SLC26A2, SLC35D1, SOX9, TBX6, TCTEX1D2, TMEM38B, TRAF3IP1, TRIP11, TRIP4, TRPV4, TTC21B, WDR19, WDR34, WDR35, WDR60, WNT1, WNT3

55002	小児 遺伝学	Invitae白質ジストロフィーと遺伝性白質脳症パネル Invitae Leukodystrophy and Genetic Leukoencephalopathy Panel	AARS, AARS2, ABAT, ABCA1, ABCD1, ABCD4, ABHD5, ACADS, ACBD5, ACER3, ACO2, ACOX1, ACP5, ACSF3, ACTA2, ACTB, ACY1, ADAR, ADGRG1, ADK, ADNP, ADSL, AGA, AHDC1, AHI1, AIFM1, AIMP1, AIMP2, ALDH18A1, ALDH3A2, ALDH5A1, ALDH6A1, ALG12, ALG13, ALG2, ALG6, ALG9, AMACR, AMPD2, AMT, ANK3, AP1S2, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APC2, APOPT1, ARCN1, ARFGEF2, ARHGAP31, ARHGEF9, ARNT2, ARSA, ARX, ASL, ASNS, ASPA, ASS1, ASXL1, ASXL2, ATP13A2, ATP5A1, ATP6AP2, ATP6V1A, ATP7A, ATP7B, ATP8A2, ATPAF2, ATRN, ATRX, AUH, B3GALNT2, B4GALNT1, BCAP31, BCAT2, BCKDHA, BCKDHB, BCL11B, BCS1L, BICD2, BMP4, BOLA3, BPTF, BRAT1, BTD, C12ORF57, C12ORF65, C19ORF12, C2CD3, CACNA1A, CACNA1E, CARS2, CBS, CC2D2A, CCDC88A, CDC42, CDKL5, CEP290, CHMP1A, CLCN2, CLCN4, CLCN7, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLP1, CLPP, CNNM2, CNOT1, CNTNAP1, CNTNAP2, COA7, COASY, COG7, COL18A1, COL3A1, COL4A1, COL4A2, COLGALT1, COQ2, COQ6, COQ7, COQ8A, COQ9, COX10, COX14, COX15, COX20, COX6B1, COX7B, COX8A, CPLANE1, CPLX1, CPS1, CRAT, CREBBP, CRIPT, CRLF1, CSF1R, CSPP1, CTBP1, CTC1, CTDP1, CTNS, CTSA, CTSD, CUL4B, CYB5R3, CYFIP2, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS, DARS2, DBT, DCAF17, DCX, DDC, DDHD1, DDHD2, DDOST, DDX3X, DEAF1, DEGS1, DGUOK, DHCR24, DHFR, DHX37, DLD, DLL4, DMXL2, DNM1L, DNM2, DOCK6, DOCK7, DOLK, DONSON, DPAGT1, DPM1, DPM2, DPM3, DPYS, DYRK1A, EARS2, EDNRB, EHMT1, EIF2AK1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL1, ELOVL4, ENTPD1, EOGT, EPG5, EPRS, ERCC1, ERCC2, ERCC3, ERCC6, ERCC8, ETFA, ETFB, ETFDH, ETHE1, EXOSC2, EXOSC3, EXOSC8, EXOSC9, FA2H, FAM126A, FAR1, FARS2, FARSB, FASTKD2, FBXL4, FDX2, FGFRL1, FH, FIG4, FKRP, FKTN, FLVCR2, FOLR1, FOXC1, FOXG1, FOXRED1, FUCA1, FUK, GAA, GABBR2, GABRA1, GABRB1, GABRB2, GABRB3, GALC, GALT, GAN, GATAD2B, GCDH, GDAP1, GFAP, GFM1, GFM2, GJA1, GJB1, GJC2, GLA, GLB1, GLDC, GLRX5, GLUL, GLYCTK, GM2A, GMNN, GNAO1, GNS, GOT2, GPHN, GRIN1, GRM7, GTF2H5, GTPBP2, HACE1, HCN1, HEPACAM, HERC1, HEXA, HEXB, HIBCH, HIKESHI, HK1, HLCS, HMGCL, HNRNPU, HSD17B10, HSD17B4, HSPD1, IARS, IARS2, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, INPP5E, IREB2, ISCA1, ISCA2, ITPA, ITPR1, IVD, JAM3, KARS, KAT6B, KATNB1, KCNJ10, KCNJ2, KCNMA1, KCNT1, KCTD7, KDM1A, KIAA0556, KIAA0586, KIAA0753, KIDINS220, KIF1A, KIF1C, KLHL7, KMT2E, L2HGDH, LAGE3, LAMA1, LAMA2, LAMB1, LARGE1, LARS2, LETM1, LIAS, LIPT1, LIPT2, LONP1, LRPPRC, LYRM7, MAG, MAGEL2, MAN2B1, MANBA, MARS2, MAST1, MAT1A, MCCC1, MCCC2, MCOLN1, MDH2, MECP2, MED17, MED25, MEF2C, MFSD2A, MFSD8, MGP, MICU1, MKS1, MLC1, MLYCD, MMACHC, MMADHC, MOCS1, MOCS2, MOGS, MORC2, MPLKIP, MPV17, MPZ, MRPL12, MRPL44, MRPS16, MRPS22, MRPS34, MSTO1, MTFMT, MTHFR, MTHFS, MTO1, MTOR, MTR, MTRR, MUT, NACC1, NADK2, NAGA, NAGLU, NAGS, NANS, NARS2, NAXD, NAXE, NDRG1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB10, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEK1, NFE2L2, NFU1, NGLY1, NKX6-2, NOTCH1, NPC1, NPC2, NPHP1, NRXN1, NSUN3, NT5C2, NTRK2, NUBPL, NUP62, OAT, OCRL, OPA1, OPA3, OSGEP, OTC, PACS1, PAFAH1B1, PAH, PANK2, PARS2, PAX1, PC, PCCA, PCCB, PCDH12, PCGF2, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGK1, PHGDH, PHYH, PIGA, PIGB, PIGG, PIGL, PIGM, PIGN, PIGP, PIGQ, PIGT, PIGU, PIGV, PIK3C2A, PIK3R2, PLA2G6, PLAA, PLEKHG2, PLP1, PMM2, PMP22, PNKP, PNPT1, POLG, POLG2, POLR1A, POLR1C, POLR3A, POLR3B, POMGNT1, POMK, POMT1, POMT2, PPP1R15B, PPP2R1A, PPP3CA, PPT1, PRF1, PRKDC, PRODH, PROSC, PRPS1, PRUNE1, PSAP, PSAT1, PSPH, PTEN, PTPN23, PURA, PUS3, PYCR2, QARS, QRSL1, RAB11B, RAB3GAP1, RAB3GAP2, RARS, RARS2, RBPJ, REPS1, RERE, RHOBTB2, RIN2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RPGRIP1L, RPIA, RPS6KC1, RRM2B, RTTN, RXYLT1, SAMD9L, SAMHD1, SCN3A, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SEPSECS, SERAC1, SETBP1, SGSH, SH3TC2, SHOC2, SHPK, SLC12A2, SLC12A5, SLC12A6, SLC13A3, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC1A2, SLC1A3, SLC1A4, SLC25A1, SLC25A12, SLC25A15, SLC25A19, SLC25A22, SLC25A4, SLC25A42, SLC25A46, SLC2A1, SLC30A10, SLC33A1, SLC35A2, SLC39A14, SLC39A8, SLC46A1, SLC6A19, SLC6A8, SLC6A9, SLC9A1, SLC9A6, SMC1A, SNAP29, SNIP1, SNORD118, SNRPB, SNX14, SOD1, SON, SOX10, SOX2, SPART, SPAST, SPATA5, SPG11, SPG7, SPTAN1, SQSTM1, SRD5A3, SSR4, ST3GAL5, STAG2, STAMBP, STAT1, STAT2, STN1, STRADA, STX11, STXBP1, STXBP2, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYNE1, SYNJ1, TACO1, TAF2, TANGO2, TARS2, TBC1D24, TBCD, TBCE, TBCK, TBX1, TCF4, TCTN2, TIMM50, TIMMDC1, TM4SF20, TMEM106B, TMEM126B, TMEM165, TMEM216, TMEM67, TMEM70, TMTC3, TOE1, TP53RK, TPI1, TPK1, TRAPPC11, TRAPPC9, TREX1, TRMT10A, TRMT5, TSC1, TSC2, TSEN54, TTC19, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUFM, TWNK, TXN2, TYMP, UBA5, UBE2A, UBE3A, UFM1, UNC13D, UPB1, USP7, VARS2, VPS11, VPS13D, VPS33A, WARS2, WDR45, WDR73, WHSC1, WWOX, YARS, YME1L1, ZEB2, ZFYVE26, ZIC1, ZNF335
4111	小児 遺伝学	Invitaeジュベール症候群およびMeckel-Gruber症候群パネル Invitae Joubert and Meckel-Gruber Syndromes Panel	AHI1, ARL13B, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, INPP5E, KIAA0586, KIF7, MKS1, MRE11, NPHP1, NPHP3, OFD1, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423

4101	小児 遺伝学	Invitae原発性線毛機能不全パネル Invitae Primary Ciliary Dyskinesia Panel	AK7, ARMC4, C11ORF70, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CEP164, CFAP298, CFTR, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, GAS8, LRRC56, LRRC6, MCIDAS, NOTCH2, OFD1, PIH1D3, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, ZMYND10
4163	小児 遺伝学	Invitae Cardio-Facio-Cutaneous Syndrome Panel	BRAF, KRAS, MAP2K1, MAP2K2, SHOC2, SOS1
4165	小児 遺伝学	Invitaeレジウス症候群検査 Invitae Legius Syndrome Test	SPRED1
1708	小児 遺伝学	Invitae神経線維腫症1型検査 Invitae Neurofibromatosis Type 1 Test	NF1
4714	小児 遺伝学	Invitae嚢胞性線維症検査 Invitae Cystic Fibrosis Test	CFTR
4721	小児 遺伝学	InvitaeATR-X症候群検査 Invitae Alpha Thalassemia X-linked Intellectual Disability Test	ATRX
4741	小児 遺伝学	InvitaeBaraitser-冬のCerebrofrontofacial症候群テスト Invitae Baraitser-Winter Cerebrofrontofacial Syndrome Test	ACTB, ACTG1
4725	小児 遺伝学	Invitaeカーペンター症候群パネル Invitae Carpenter Syndrome Panel	MEGF8, RAB23
4738	小児 遺伝学	Invitaeコフィン・ローリー症候群検査 Invitae Coffin-Lowry Syndrome Test	RPS6KA3
4737	小児 遺伝学	Invitaeコーエン症候群検査 Invitae Cohen Syndrome Test	VPS13B
4727	小児 遺伝学	Invitaeコルネリア・デランゲ症候群および関連障害パネル Invitae Cornelia de Lange Syndrome and Related Disorders Panel	ADNP, AFF4, ANKRD11, ARID1A, ARID1B, BRD4, CREBBP, DPF2, EP300, ESCO2, HDAC8, KMT2A, MED13L, NIPBL, PHF6, PHIP, RAD21, SETD5, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SOX11, SRCAP, STAG1, STAG2, TAF1, TAF6, ZMYND11
4303	小児 遺伝学	Invitae顔面異骨症および前鼻異形成パネル Invitae Facial Dysostosis and Frontonasal Dysplasia Panel	ALX1, ALX3, ALX4, CHD7, DHODH, EDN1, EDNRA, EFNB1, EFTUD2, EVC, EVC2, GATA1, GNAI3, IRX5, PDE4D, PLCB4, POLR1A, POLR1C, POLR1D, PRKAR1A, RPL11, RPL5, RPS28, SF3B4, TCOF1, TSR2, TWIST1, ZSWIM6
4736	小児 遺伝学	Invitae低ゴナドトロピン性性腺機能低下症パネル Invitae Hypogonadotropic Hypogonadism Panel	ANOS1, AXL, CCDC141, CHD4, CHD7, CYP19A1, DHCR7, DUSP6, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IGSF10, IL17RD, KISS1, KISS1R, KLB, LEP, LEPR, LHB, LMNA, NR0B1, NSMF, PCSK1, PLXNA1, POLR3B, PROK2, PROKR2, PROP1, RELN, RNF216, SEMA3A, SOX10, SOX2, SPRY4, SRA1, SRY, TAC3, TACR3, WDR11
4747	小児 遺伝学	InvitaeKBG症候群検査 Invitae KBG Syndrome Test	ANKRD11
4213	小児 遺伝学	InvitaeOculo-Facio-Cardio-Dental Syndrome Test	BCOR

4735	小児 遺伝学	Invitae vanderWoude症候群パネル Invitae van der Woude Syndrome Panel	GRHL3, IRF6
5021	小児 遺伝学	Invitae外胚葉異形成症（歯の無形成の有無を問わない）パネル Invitae Ectodermal Dysplasia with or without Tooth Agenesis Panel	ANTXR1, APCDD1, ATP7A, AXIN2, BCS1L, BMP4, CDH3, CDSN, CLDN1, CTSC, DSG4, DSP, EDA, EDAR, EDARADD, EGFR, ERCC2, ERCC3, ERCC8, GJA1, GJB2, GJB6, GRHL2, GTF2E2, GTF2H5, HOXC13, HR, IRF6, JUP, KANK2, KDF1, KREMEN1, KRT14, KRT16, KRT17, KRT25, KRT6A, KRT6B, KRT6C, KRT71, KRT74, KRT83, KRT85, LIPH, LPAR6, LRP6, LSS, LTBP3, MBTPS2, MPLKIP, MSX1, NECTIN1, NECTIN4, NFKBIA, PAX9, PKP1, POC1A, PORCN, PTH1R, RIN2, RNF113A, RPL21, SMOC2, SNRPE, SOX18, SPINK5, TFAP2B, TP63, TRPS1, TRPV3, TSPEAR, WNT10A, WNT10B
4304	小児 遺伝学	Invitae低色素沈着パネル Invitae Hypopigmentation Panel	ACD, AP3B1, AP3D1, BLOC1S3, BLOC1S6, C10ORF11, CLCN7, CTC1, DKC1, DTNBP1, EDN3, EDNRB, EPG5, FRMD7, GNAI3, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, KIT, KITLG, LYST, MITF, MLPH, MYO5A, NHP2, NOP10, OCA2, PARN, PAX3, RAB27A, RET, RTEL1, SLC24A5, SLC38A8, SLC45A2, SNAI2, SOX10, TERC, TERT, TINF2, TYR, TYRP1, USB1, WRAP53
434348	小児 遺伝学	Invitae眼皮膚白皮症パネル Invitae Oculocutaneous Albinism Panel	AP3B1, AP3D1, BLOC1S3, BLOC1S6, C10ORF11, DTNBP1, FRMD7, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, MLPH, MYO5A, OCA2, RAB27A, SLC24A5, SLC38A8, SLC45A2, TYR, TYRP1
434351	小児 遺伝学	Invitae色素性乾皮症パネル Invitae Xeroderma Pigmentosum Panel	DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC
4305	小児 遺伝学	Invitae色覚異常パネル Invitae Achromatopsia Panel	ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H, RGS9, RGS9BP
5132	小児 遺伝学	Invitae白内障パネル Invitae Cataracts Panel	ABCA3, ABCB6, ABHD12, ADAMTS18, ADAMTSL4, AGK, ALDH18A1, BCOR, BEST1, BFSP1, BFSP2, CHMP4B, CLN3, COL11A1, COL18A1, COL2A1, COL4A1, COL4A2, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, CYP27A1, CYP51A1, EPG5, EPHA2, ERCC2, ERCC5, ERCC6, ERCC8, EYA1, FAM126A, FOXC1, FOXE3, FTL, FYCO1, FZD4, GALK1, GALT, GCNT2, GFER, GJA1, GJA3, GJA8, HMX1, HSF4, JAM3, LEMD2, LIM2, LONP1, LSS, MAF, MIP, MIR184, MYH9, NDP, NF2, NHS, OCRL, OPA3, P3H2, PAX6, PEX10, PEX11B, PEX16, PEX2, PEX7, PITX2, PITX3, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RDH11, RECQL4, RGS6, RNLS, RRAGA, SC5D, SIL1, SIPA1L3, SIX6, SLC16A12, SLC33A1, TBC1D20, TDRD7, TFAP2A, TMEM70, UNC45B, VIM, VSX2, WDR87, WFS1, WRN, XYLT2
4301	小児 遺伝学	Invitae先天性静止夜盲目パネル Invitae Congenital Stationary Night Blindness Panel	CABP4, CACNA1F, CACNA2D4, CHM, CYP4V2, FRMD7, GNAT1, GNB3, GPR179, GRM6, GUCY2D, LRIT3, NYX, PDE6B, RBP4, RDH5, RHO, RLBP1, RPE65, SAG, SLC24A1, TRPM1
4302	小児 遺伝学	Invitae角膜ジストロフィパネル Invitae Corneal Dystrophies Panel	CHRDL1, CHST6, COL17A1, COL5A1, COL8A2, CYP4V2, DCN, FOXE3, GJA8, GRHL2, GSN, KERA, KRT12, KRT3, LCAT, LOXHD1, MAF, MIR184, NLRP1, OVOL2, PAX6, PIKFYVE, PITX2, PRDM5, PXDN, SLC4A11, TACSTD2, TGFBI, UBIAD1, VSX1, ZEB1, ZNF143, ZNF469
55015	小児 遺伝学	Invitae緑内障パネル Invitae Glaucoma Panel	ASB10, ATOH7, BMP4, COL4A1, COL8A2, CYP1B1, EXO5, FOXC1, FOXE3, LMX1B, LTBP2, MAF, MFRP, MYOC, OPTN, PAX6, PIK3R1, PITX2, PITX3, PRPF8, PRSS56, PXDN, SH3PXD2B, SIX6, SLC4A4, TEK, WDR36
434345	小児 遺伝学	InvitaeMacularジストロフィーパネル Invitae Macular Dystrophy Panel	ABCA4, BEST1, C1QTNF5, CDH3, CERKL, CFI, CHST6, CNGB3, CRB1, CRX, CTNNA1, DRAM2, EFEMP1, ELOVL4, FSCN2, GUCA1B, HMCN1, IMPG1, IMPG2, KCNV2, MFSD8, NMNAT1, PRDM13, PROM1, PRPH2, RAX2, RBP3, RBP4, RDH12, RDH5, RLBP1, RP1L1, RPGRIP1, RS1, SIX6, TIMP3

72100	小児 遺伝学	Invitae継承網膜障害パネル Invitae Inherited Retinal Disorders Panel	ABCA4, ABCC6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADAMTSL4, ADGRA3, ADGRV1, ADIPOR1, AGBL5, AHI1, AHR, AIPL1, ALMS1, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARSG, ASRGL1, ATF6, ATOH7, B9D1, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C10ORF11, C12ORF65, C1QTNF5, C8ORF37, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CCT2, CDH23, CDH3, CDHR1, CEP164, CEP19, CEP250, CEP290, CEP41, CEP78, CEP83, CERKL, CFAP410, CHM, CIB2, CISD2, CLCC1, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLRN1, CLUAP1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, CPLANE1, CRB1, CRX, CSPP1, CTNNA1, CTSD, CWC27, CYP4V2, DHDDS, DHX32, DHX38, DNAJC17, DRAM2, DSCAML1, DTHD1, EFEMP1, ELOVL4, EMC1, ERCC6, EXOSC2, EYS, FAM161A, FBLN5, FLVCR1, FRMD7, FSCN2, FZD4, GDF6, GNAT1, GNAT2, GNB3, GNPTG, GNS, GPR143, GPR179, GPR45, GRM6, GRN, GUCA1A, GUCA1B, GUCY2D, HARS, HGSNAT, HK1, HMCN1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT43, IFT74, IFT80, IFT81, IFT88, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, ITM2B, JAG1, KCNJ13, KCNV2, KIAA0586, KIAA1549, KIF11, KIF7, KIZ, KLHL7, LCA5, LRAT, LRIT3, LRP2, LRP5, LYST, LZTFL1, MAK, MAPKAPK3, MERTK, MFN2, MFRP, MFSD8, MIR204, MKKS, MKS1, MPDZ, MTPAP, MTTP, MYO7A, NAGLU, NBAS, NDP, NEK2, NEUROD1, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OCA2, OFD1, OPA1, OPA3, OPN1SW, OR2W3, OTX2, P3H2, PAX2, PAX6, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PITPNM3, PLA2G5, PLK4, PNPLA6, POC1B, POC5, POMGNT1, PPT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RAB28, RAX2, RBP1, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGR (ORF15), RPGRIP1, RPGRIP1L, RS1, RTN4IP1, SAG, SAMD11, SCLT1, SDCCAG8, SEMA4A, SGSH, SIX6, SLC24A1, SLC24A5, SLC45A2, SLC7A14, SNRNP200, SPATA7, SPP2, TCTN1, TCTN2, TCTN3, TEAD1, TIMM8A, TIMP3, TMED7, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TRAF3IP1, TREX1, TRIM32, TRNT1, TRPM1, TSPAN12, TTC21B, TTC8, TTLL5, TTPA, TUB, TUBGCP4, TUBGCP6, TULP1, TYR, TYRP1, UNC119, USH1C, USH1G, USH2A, VCAN, VPS13B, WDPCP, WDR19, WDR34, WFS1, WHRN, ZNF408, ZNF423, ZNF513
4201	小児 遺伝学	Invitae先天性心疾患および内臓錯位パネル Invitae Congenital Heart Defects and Heterotaxy Panel	ACTC1, ACVR2B, ALMS1, ANKS6, ARMC4, BBS10, BCOR, BRAF, CBL, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CEP290, CFAP298, CFAP53, CHD7, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAI1, DNAI2, DNAL1, DRC1, ELN, FOXH1, GAS8, GATA4, GDF1, GJA1, GPC3, HRAS, INVS, JAG1, KRAS, LEFTY2, LRRC6, MAP2K1, MAP2K2, MCIDAS, MED13L, MEIS2, MKS1, NEK8, NF1, NKX2-5, NKX2-6, NME8, NODAL, NOTCH1, NOTCH2, NPHP3, NR2F2, NRAS, NSD1, OFD1, PTPN11, RAF1, RIT1, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SHOC2, SOS1, SPAG1, TBX1, TBX5, TTC8, ZIC3, ZMYND10, ZNF423
4412	小児 遺伝学	Invitaeアンドロゲン不応症パネル Invitae Androgen Insensitivity Panel	AR, SRD5A2
4413	小児 遺伝学	Invitae女性の性発達障害検査 Invitae Disorders of Female Sex Development Test	SRY
55007	小児 遺伝学	Invitae性発達障害パネル Invitae Disorders of Sex Development Panel	AMH, AMHR2, ANOS1, AR, ARX, ATRX, B3GLCT, CBX2, CCNQ, CHD7, CKAP2L, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, DHH, DMRT1, ESR2, FRAS1, FREM2, H6PD, HHAT, HOXA13, HSD17B3, HSD3B2, KL, LHCGR, LHX3, LHX4, MAMLD1, MAP3K1, NR0B1, NR5A1, POR, PROP1, PSMC3IP, RSPO1, SOX9, SPECC1L, SRD5A2, SRY, STAR, TNK2, TOE1, TSPYL1, TWIST2, UBR1, WNT4, WNT9B, WT1, WWOX, ZFPM2
434342	小児 遺伝学	Invitaeアダムズ・オリバー症候群パネル Invitae Adams-Oliver Syndrome Panel	ARHGAP31, DLL4, DOCK6, EOGT, KCTD1, NOTCH1, RBPJ, UBR1
4726	小児 遺伝学	Invitae ARSE関連の点状軟骨異形成症検査 Invitae ARSE-Related Chondrodysplasia Punctata Test	ARSE
4712	小児 遺伝学	Invitae屈曲肢異形成症検査 Invitae Campomelic Dysplasia Test	SOX9

4423	小児 遺伝学	Invitae頭蓋骨癒合症パネル Invitae Craniosynostosis Panel	ALPL, ASXL1, B3GAT3, CD96, CDC45, CDT1, COLEC11, CYP26B1, EFNA4, EFNB1, ERF, ESCO2, FBN1, FGF9, FGFR1, FGFR2, FGFR3, FREM1, GLI3, GPC3, IFT122, IFT140, IFT43, IGF1R, IL11RA, KAT6A, KAT6B, MASP1, MEGF8, MSX2, NFIA, ORC1, ORC4, ORC6, P4HB, PHEX, POR, PPP3CA, RAB23, RECQL4, RSPRY1, RUNX2, SCARF2, SEC24D, SIX2, SKI, SLC25A24, SMAD2, SMAD3, SMAD6, SOX6, SPECC1L, STAT3, TCF12, TCOF1, TGFB2, TGFB3, TGFBR1, TGFBR2, TMCO1, TWIST1, WDR19, WDR35, ZEB2, ZIC1
4613	小児 遺伝学	Invitaeエリス・ファンクレフェルトおよびWeyers四肢顔面骨形成不全症パネル Invitae Ellis van Creveld and Weyers Acrofacial Dysostosis Panel	EVC, EVC2
4614	小児 遺伝学	Invitae遺伝性多発性骨軟骨腫パネル Invitae Hereditary Multiple Osteochondromas Panel	EXT1, EXT2
72039	小児 遺伝学	Invitae低リン血症パネル Invitae Hypophosphatemia Panel	ALPL, CLCN5, CTNS, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FAM20C, FGF23, FGFR1, GNAS, OCRL, PHEX, SLC34A1, SLC34A3, VDR
55010	小児 遺伝学	Invitae指趾の奇形パネル Invitae Limb and Digital Malformations Panel	ACVR1, ADAMTS10, ADAMTS17, AFF4, AHI1, ANKRD11, ARHGAP31, ARID1A, ARID1B, ARL13B, ARL6, B3GLCT, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BHLHA9, BMP2, BMP4, BMPR1B, BTRC, C2CD3, CACNA1C, CC2D2A, CCNQ, CDH3, CEP104, CEP120, CEP290, CEP41, CHSY1, CHUK, CKAP2L, CPLANE1, CREBBP, CSPP1, DDX59, DHCR7, DHODH, DLL4, DLX5, DLX6, DOCK6, DPF2, DVL1, DVL3, DYNC1I1, EOGT, EP300, ESCO2, EVC, EVC2, FAT1, FBLN1, FBN1, FBXW4, FGF10, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FLNA, FMN1, FRAS1, FREM2, FZD2, GDF5, GDF6, GJA1, GLI2, GLI3, GNAS, GSC, HDAC4, HDAC8, HOXA13, HOXD13, IFT57, IHH, INPP5E, KDM6A, KIAA0586, KIF7, KMT2A, KMT2D, LMBR1, LRP4, LTBP2, LTBP3, MAP3K20, MEGF8, MGP, MKKS, MKS1, MRE11, MYCN, NECTIN1, NECTIN4, NIPBL, NOG, NOTCH1, NPHP1, NPHP3, NSDHL, NXN, OFD1, PDE3A, PDE4D, PDE6D, PGM3, PHF6, PIGV, PITX1, POLR1A, PORCN, PRKAR1A, PRMT7, PTDSS1, PTHLH, RAB23, RAD21, RBM8A, RBPJ, RECQL4, ROR2, RPGRIP1L, SALL1, SALL4, SC5D, SDCCAG8, SF3B4, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SMOC1, SOST, SOX11, SOX9, TBX15, TBX3, TBX5, TCTN1, TCTN2, TCTN3, TGDS, THPO, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TP63, TRIM32, TRPS1, TRPV4, TTC21B, TTC8, VAC14, WDPCP, WNT10B, WNT3, WNT5A, WNT7A, ZNF423, ZSWIM6
4307	小児 遺伝学	Invitae小頭症原発性小人症およびセッケル症候群パネル Invitae Microcephalic Primordial Dwarfism and Seckel Syndrome Panel	ATR, ATRIP, CDC45, CDC6, CDK5RAP2, CDT1, CENPJ, CEP135, CEP152, CEP63, CEP97, CRIPT, DNA2, DNMT3A, DONSON, GMNN, LARP7, LIG4, MCM5, MCPH1, NIN, NSMCE2, ORC1, ORC4, ORC6, PCNT, PLK4, POC1A, RBBP8, RNU4ATAC, RTTN, SRCAP, TRAIP, TUBGCP4, TUBGCP6, UBE3B, WDR4, XRCC4
434350	小児 遺伝学	Invitae脊椎肋骨異骨症パネル Invitae Spondylocostal Dysostosis Panel	DLL1, DLL3, DMRT2, HES7, LFNG, MESP2, RIPPLY2, TBX6
4617	小児 遺伝学	Invitae毛髪-鼻-指節症候群パネル Invitae Trichorhinophalangeal Syndrome Panel	EXT1, TRPS1
72038	小児 遺伝学	InvitaeX連鎖性低リン血症性検査 Invitae X-Linked Hypophosphatemia Test	PHEX

4306	小児 遺伝学	Invitae界面活性剤代謝パネル Invitae Surfactant Metabolism Panel	ABCA3, COPA, CSF2RA, CSF2RB, DKC1, FLNA, FOXF1, GATA2, ITGA3, MARS, NKX2-1, NOTCH2, PARN, RTEL1, SFTPB, SFTPC, SLC7A7, TERC, TERT, TINF2, TMEM173
55014	小児 遺伝学	Invitaeアッシャー症候群パネル Invitae Usher Syndrome Panel	ABHD12, ADGRV1, ARSG, CDH23, CEP250, CIB2, CLRN1, HARS, MYO7A, PCDH15, PDZD7, PEX1, PEX6, USH1C, USH1G, USH2A, WHRN
434341	小児 遺伝学	腎臓および尿路のInvitae先天性異常（CAKUT）パネル Invitae Congenital Anomalies of Kidney and Urinary Tract (CAKUT) Panel	ACE, AGT, AGTR1, BICC1, BNC2, CRKL, DCHS1, DSTYK, EYA1, FAT4, FGF20, FOXC1, FRAS1, FREM1, FREM2, GATA3, GLI3, GREB1L, HNF1B, HOXA13, HPSE2, ITGA8, LRP4, NRIP1, PAX2, PBX1, REN, RET, ROBO2, SALL1, SIX1, SIX2, SIX5, SLIT2, SOX17, TBX18, UMOD, VIPAS39, VPS33B, WNT4, WT1
55008	小児 遺伝学	Invitae嚢胞性腎疾患パネル Invitae Cystic Kidney Disease Panel	ALG8, ANKS6, BICC1, CEP164, CEP290, CEP83, CEP89, COL4A1, CRB2, DCDC2, DICER1, DNAJB11, DZIP1L, GANAB, GLIS2, HNF1B, IFT172, INVS, IQCB1, JAG1, LRP5, MAPKBP1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD2, PKHD1, PRKCSH, RPGRIP1L, SDCCAG8, SEC61A1, SEC63, TMEM67, TSC1, TSC2, TTC21B, UMOD, VHL, WDR19, ZNF423
434347	小児 遺伝学	Invitaeネフローゼ症候群および巣状分節性糸球体硬化症(FSGS)パネル Invitae Nephrotic Syndrome and Focal Segmental Glomerulosclerosis (FSGS) Panel	ACTN4, AMN, ANLN, APOL1, ARHGAP24, ARHGDIA, CD2AP, CFH, CLCN5, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CUBN, DGKE, DHTKD1, EMP2, FN1, INF2, ITGA3, ITGB4, KANK1, KANK2, KANK4, LAGE3, LAMA5, LAMB2, LMX1B, MAGI2, MYO1E, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, NXF5, OCRL, OSGEP, PAX2, PDSS2, PLCE1, PODXL, PTPRO, SGPL1, SMARCAL1, TP53RK, TPRKB, TRPC6, WDR73, WT1, XPO5
75000	小児 遺伝学	Invitae進行性腎疾患パネル Invitae Progressive Renal Disease Panel	ACE, ACTB, ACTN4, ADAMTS13, AGT, AGTR1, AHI1, ALG8, ALG9, ALMS1, AMN, ANKS6, ANLN, APOL1, APRT, AQP2, ARHGAP24, ARHGDIA, ARL6, ATP6V0A4, ATP6V1B1, AVP, AVPR2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BICC1, BSND, CA2, CACNA1D, CACNA1H, CASR, CD151, CD2AP, CDKN1C, CEP164, CEP19, CEP290, CEP83, CEP89, CFHR5, CHRM3, CLCN5, CLCNKB, CLDN16, CLDN19, COL4A1, COL4A3, COL4A4, COL4A5, COL4A6, COQ2, COQ6, COQ8B, CPLANE1, CRB2, CTNS, CUBN, CYP24A1, DCDC2, DGKE, DHCR7, DICER1, DLC1, DNAJB11, DZIP1L, EGF, EMP2, EYA1, FAM20A, FAN1, FAT1, FN1, FOXI1, FOXP3, FREM1, GANAB, GATA3, GATM, GLA, GLIS2, GRHPR, GSN, HNF1A, HNF1B, HPRT1, HPSE2, HSD11B2, IFT122, IFT140, IFT172, IFT27, IFT43, INF2, INVS, IQCB1, ITGA3, ITGA6, ITGB4, JAG1, KANK2, KANK4, KAT6B, KCNJ1, KCNJ10, LAMA5, LAMB2, LCAT, LDHA, LMX1B, LPIN1, LRP4, LRP5, LYZ, LZTFL1, MAGED2, MAGI2, MAPKBP1, MEFV, MKKS, MYH9, MYO1E, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NSDHL, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, OFD1, PAX2, PBX1, PDSS2, PHEX, PKD2, PKHD1, PLCE1, PODXL, PREPL, PTPRO, REN, RMND1, RPGRIP1L, SALL1, SALL4, SARS2, SDCCAG8, SEC61A1, SEMA3E, SGPL1, SIX1, SIX5, SLC12A1, SLC12A3, SLC22A12, SLC2A2, SLC2A9, SLC37A4, SLC41A1, SLC4A1, SLC4A4, SLC7A7, SMARCAL1, SYNPO, TBC1D8B, TMEM67, TNS2, TP63, TRIM32, TRPC6, TSC1, TSC2, TTC21B, TTC8, UMOD, WDPCP, WDR19, WDR73, WT1, XPNPEP3, XPO5, ZNF423
4308	小児 遺伝学	Invitae腎尿細管性障害パネル Invitae Renal Tubular Disorders Panel	ACE, AGT, AGTR1, AQP2, ATP6V0A4, ATP6V1B1, AVPR2, BSND, CA2, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, CUL3, EGF, FOXI1, FXYD2, GATM, GNA11, HSD11B2, KCNJ1, KCNJ10, KLHL3, MAGED2, NR3C2, OCRL, REN, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A3, SLC34A1, SLC4A1, SLC4A4, TRPM6, WNK1, WNK4
4167	小児 遺伝学	Invitae神経線維腫症2型検査 Invitae Neurofibromatosis Type 2 Test	NF2

4422	小児 遺伝学	Invitae脳海綿状血管腫パネル Invitae Cerebral Cavernous Malformations Panel	CCM2, KRIT1, PDCD10
55004	小児 遺伝学	Invitae脳性麻痺スペクトラム障害パネル Invitae Cerebral Palsy Spectrum Disorders Panel	ABAT, ABCD1, ACADM, ACADVL, ACAT1, ACBD5, ACOX1, ACTB, ADAR, ADCY5, ADD3, ADNP, ADSL, AFG3L2, AGAP1, AHDC1, AHI1, AKT3, ALDH18A1, ALDH3A2, ALDH5A1, ALDH7A1, ALG13, ALG3, ALS2, AMACR, AMPD2, AMT, ANO3, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APTX, ARG1, ARHGEF9, ARL6IP1, ARSA, ARX, ASL, ASNS, ASPA, ASS1, ASXL1, ATAD1, ATL1, ATM, ATP13A2, ATP1A3, ATP7A, ATP7B, ATP8A2, ATRX, AUH, AUTS2, B4GALNT1, BCAP31, BCKDHA, BCKDHB, BICD2, BSCL2, BTD, C12ORF65, C19ORF12, CACNA1A, CACNA1G, CAMTA1, CAPN1, CASK, CBS, CCDC88C, CCT5, CDKL5, CEP290, CHD8, CHRNA1, CIZ1, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLTC, COASY, COL4A1, COL4A2, COL6A3, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ9, CPS1, CPT1C, CREBBP, CTBP1, CTNNB1, CTSD, CYP27A1, CYP2U1, CYP7B1, DARS, DARS2, DBH, DBT, DCAF17, DDC, DDHD1, DDHD2, DDX3X, DGKZ, DHDDS, DHFR, DLAT, DLD, DMD, DNAJC12, DNM2, DPAGT1, DYNC1H1, DYRK1A, EEF2, EHMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, ENTPD1, EPHA4, ERCC6, ERCC8, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, FA2H, FAM126A, FARS2, FAT2, FGF12, FGF14, FH, FOLR1, FOXG1, FRRS1L, FTL, FUCA1, GABRA2, GAD1, GALC, GAMT, GATM, GBA, GBA2, GCDH, GCH1, GFAP, GJC2, GLB1, GLDC, GLRA1, GLRB, GM2A, GNAL, GNAO1, GNB1, GNS, GPHN, GPR88, GRID2, GRIN1, GRIN2B, GRM1, HACE1, HESX1, HEXA, HEXB, HGSNAT, HLCS, HMGCL, HPCA, HPRT1, HSD17B10, HSD17B4, HSPD1, IBA57, IFIH1, IQSEC2, IREB2, ITPA, ITPR1, KANK1, KAT6A, KCNA2, KCNC3, KCNJ6, KCNMA1, KCNQ2, KCNT1, KCTD17, KCTD7, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, KMT2A, KMT2B, KMT2C, L1CAM, L2HGDH, LAMA2, LIAS, LMBRD1, MAG, MAOA, MAP2K1, MARS2, MAST1, MCCC1, MCCC2, MCEE, MECP2, MECR, MFSD8, MICU1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MOCS2, MOCS3, MPC1, MTHFR, MTOR, MTPAP, MTR, MTRR, MTTP, MUT, NAA10, NAA35, NAGLU, NAGS, NBAS, NGLY1, NIPA1, NKX2-1, NPC1, NPC2, NPHP1, NT5C2, NUS1, OTC, PAFAH1B1, PAH, PAK3, PALM, PANK2, PCBD1, PCCA, PCCB, PCDH12, PDE10A, PDE2A, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDYN, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PHGDH, PHIP, PIGN, PIGT, PLA2G6, PLD3, PLP1, PLXNA2, PMM2, PNKD, PNP, PNPLA6, PNPO, POLG, POLR3A, PPT1, PRKRA, PROSC, PRRT2, PRUNE1, PSAT1, PSPH, PTPN11, PTS, PURA, QDPR, RAB3GAP1, RAB3GAP2, RANBP2, REEP1, REEP2, RHOBTB2, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RTN2, SACS, SAMHD1, SATB2, SCN1A, SCN2A, SCN3A, SCN8A, SETD5, SGCE, SGSH, SHH, SIL1, SIX3, SLC16A2, SLC17A5, SLC18A2, SLC19A3, SLC1A4, SLC25A15, SLC25A22, SLC2A1, SLC30A10, SLC33A1, SLC39A14, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SON, SPART, SPAST, SPATA5, SPG11, SPG21, SPG7, SPR, SPTAN1, SPTBN2, SQSTM1, ST3GAL5, STAMBP, STUB1, STXBP1, SUCLA2, SUCLG1, SUOX, SURF1, SYNGAP1, TAF1, TANGO2, TBC1D24, TBCK, TBL1XR1, TCF4, TECPR2, TFG, TGIF1, TGM6, TH, THAP1, TMEM240, TMEM67, TOR1A, TREX1, TRPC3, TSEN54, TTBK2, TTPA, TUBA1A, TUBB2A, TUBB2B, TUBB3, TUBB4A, UBE3A, UCHL1, VAC14, VAMP1, VPS13A, VPS13D, VPS37A, WARS2, WASHC5, WDR45, WDR62, ZBTB18, ZC4H2, ZEB2, ZFR, ZFYVE26, ZIC1, ZIC2, ZIC4
55009	小児 遺伝学	Inviate包括的な難聴パネル Invitae Comprehensive Deafness Panel	ABHD12, ABHD5, ACOX1, ACTG1, ADCY1, ADGRV1, AIFM1, ALMS1, ANLN, ARSB, ARSG, ATP1A3, ATP2B2, ATP6V1B1, BCAP31, BCS1L, BSND, BTD, CABP2, CACNA1D, CCDC50, CD151, CD164, CDC14A, CDH23, CEACAM16, CEP250, CEP78, CHD7, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DBH, DCAF17, DCDC2, DFNA5, DFNB59, DIABLO, DIAPH1, DIAPH3, DMXL2, DNMT1, DSPP, EDN3, EDNRA, EDNRB, EFTUD2, ELMOD3, EPS8, EPS8L2, ESPN, ESRRB, EYA1, EYA4, FAM65B, FGF3, FGFR3, FOXC1, FOXI1, GALNS, GATA3, GDF6, GIPC3, GJA1, GJB1, GJB2, GJB3, GJB6, GLB1, GNS, GPSM2, GRHL2, GRXCR1, GRXCR2, GUSB, HARS, HARS2, HGF, HGSNAT, HOMER2, HSD17B4, HYAL1, IDS, IDUA, ILDR1, JAG1, KARS, KCNE1, KCNJ10, KCNQ1, KCNQ4, KITLG, LARS2, LHFPL5, LHX3, LOXHD1, LOXL3, LRP2, LRTOMT, MAN2B1, MARVELD2, MCM2, MEOX1, MET, MIR96, MITF, MPZ, MSRB3, MYH14, MYH7B, MYH9, MYO15A, MYO18B, MYO3A, MYO6, MYO7A, NAGLU, NARS2, NDRG1, NF2, NLRP3, NOG, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PCGF2, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PMP22, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, RAI1, RDX, RMND1, ROR1, RPS6KA3, S1PR2, SCP2, SERAC1, SERPINB6, SGSH, SH3TC2, SIX1, SIX5, SLC12A2, SLC17A8, SLC22A4, SLC26A4, SLC26A5, SLC29A3, SLC44A4, SLC4A11, SLC52A2, SLC52A3, SLITRK6, SMPX, SNAI2, SOX10, SYNE4, TBC1D24, TCOF1, TECTA, TFAP2A, TIMM8A, TJP2, TMC1, TMEM126A, TMEM132E, TMIE, TMPRSS3, TPRN, TRIOBP, TRRAP, TSPEAR, TUBB4B, TWNK, UBR1, USH1C, USH1G, USH2A, VCAN, WBP2, WFS1, WHRN, ZNF469
4732	小児 遺伝学	Invitae骨形成不全症および骨脆弱性パネル Invitae Osteogenesis Imperfecta and Bone Fragility Panel	ALPL, ANO5, ASCC1, B3GAT3, B4GALT7, BMP1, CA2, CLCN5, CLCN7, COL1A1, COL1A2, CREB3L1, CRTAP, CTNS, CTSK, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FAM20C, FAM46A, FGF23, FGFR1, FKBP10, GNAS, GORAB, IFITM5, LRP5, LRRK1, MBTPS2, MESDC2, NBAS, NOTCH2, NTRK1, OCRL, OSTM1, P3H1, P4HB, PHEX, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SFRP4, SGMS2, SLC29A3, SLC2A2, SLC34A1, SLC34A3, SNX10, SP7, SPARC, SUCO, TAPT1, TCIRG1, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TRIP4, VDR, WNT1, WNT3A, XYLT2

89100	小児 遺伝学	Invitae骨系統疾患パネル Invitae Skeletal Disorders Panel	ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS17, AFF4, AGA, AGPS, AIFM1, ALPL, AMER1, ANKH, ANO5, ARCN1, ARSB, ARSE, ASCC1, ASPM, ATR, ATRIP, B3GALT6, B3GAT3, B4GALT7, BGN, BMP1, BMP2, BMPER, BMPR1B, C2CD3, CA2, CANT1, CASR, CCDC8, CDC45, CDC6, CDK5RAP2, CDKN1C, CDT1, CENPJ, CEP120, CEP135, CEP152, CEP63, CEP97, CFAP410, CHST14, CHST3, CHUK, CLCN5, CLCN7, COG1, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL27A1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CREB3L1, CRIPT, CRTAP, CSF1R, CSGALNACT1, CSPP1, CTNS, CTSA, CTSK, CUL7, CWC27, CYP27B1, CYP2R1, DDR2, DDRGK1, DHCR24, DIP2C, DLL1, DLL3, DLX3, DMP1, DMRT2, DNA2, DNMT3A, DONSON, DVL1, DVL3, DYM, DYNC2H1, DYNC2LI1, EBP, EIF2AK3, ENPP1, ESCO2, EVC, EVC2, EXOC6B, EXOSC2, EXT1, EXT2, EXTL3, FAH, FAM111A, FAM20C, FAM46A, FAR1, FAT4, FBN1, FGF23, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FLNA, FLNB, FN1, FTO, FUCA1, FZD2, GALNS, GALNT3, GDF5, GDF6, GHR, GHRHR, GHSR, GJA1, GLB1, GMNN, GNAS, GNE, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC6, GPX4, GSC, GUSB, GZF1, HES7, HGSNAT, HPGD, HSPG2, HYAL1, IARS2, ICK, IDS, IDUA, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT57, IFT74, IFT80, IFT81, IGF1, IGF2, IHH, IMPAD1, INPPL1, INTU, JAG1, KAT6B, KIAA0586, KIAA0753, KIF22, KL, KMT2A, LARP7, LBR, LEMD3, LFNG, LIFR, LIG4, LMNA, LMX1B, LONP1, LOXL3, LRP4, LRP5, LRRK1, LTBP2, LTBP3, MAFB, MAN2B1, MANBA, MAP3K7, MATN3, MBTPS1, MBTPS2, MCM5, MCPH1, MEOX1, MESDC2, MESP2, MGP, MMP13, MMP14, MMP2, MMP9, MNX1, MSX2, MYH3, MYO18B, NAGLU, NANS, NBAS, NEK1, NEU1, NIN, NKX3-2, NOG, NOTCH2, NPPC, NPR2, NPR3, NSDHL, NSMCE2, NTRK1, NXN, OBSL1, OCRL, ORC1, ORC4, ORC6, OSTM1, P3H1, P4HB, PAM16, PAPSS2, PCGF2, PCNT, PCYT1A, PDE4D, PEX5, PEX7, PGM3, PHEX, PIK3C2A, PISD, PKDCC, PLK4, PLOD2, PLS3, POC1A, POLR1A, POP1, POR, PPIB, PPP3CA, PRKAR1A, PTDSS1, PTH1R, PTHLH, PTPN11, PYCR1, RAB33B, RBBP8, RECQL4, RIPPLY2, RMRP, RNU4ATAC, ROR2, RSPO2, RSPRY1, RTTN, RUNX2, SC5D, SEC24D, SERPINF1, SERPINH1, SETBP1, SFRP4, SGMS2, SGSH, SH3PXD2B, SLC17A5, SLC26A2, SLC29A3, SLC2A2, SLC34A1, SLC34A3, SLC35D1, SLC39A13, SLCO2A1, SLCO5A1, SMAD4, SMARCAL1, SNRPB, SNX10, SOX9, SP7, SPARC, SQSTM1, SRCAP, SUCO, SULF1, SUMF1, TAB2, TAPT1, TBCE, TBX15, TBX3, TBX5, TBX6, TBXAS1, TCIRG1, TCTEX1D2, TCTN3, TGFB1, TMEM165, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TRAF3IP1, TRAIP, TRAPPC2, TREM2, TRIM37, TRIP11, TRIP4, TRMT10A, TRPS1, TRPV4, TTC21B, TUBGCP4, TUBGCP6, TYROBP, UBE3B, VAC14, VDR, VPS33A, WDR19, WDR34, WDR35, WDR4, WDR60, WISP3, WNT1, WNT3, WNT3A, WNT5A, XRCC4, XYLT1, XYLT2, ZMPSTE24, ZNF687
434340	小児 遺伝学	Invitae結合組織疾患パネル Invitae Connective Tissue Disorders Panel	ABCC6, ABL1, ACTA2, ACVR1, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL4, AEBP1, ALDH18A1, ARIH1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B3GAT3, B4GALT7, BGN, C1S, CBS, CHST14, CHST3, COG7, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, CRTAP, DCHS1, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLCN, FLNA, FLNB, FOXE3, GGCX, GORAB, HCN4, LEMD3, LOX, LOXL3, LTBP2, LTBP3, LTBP4, LZTS1, MAT2A, MED12, MFAP5, MYH11, MYLK, NOG, NOTCH1, P3H1, PKD2, PLOD1, PLOD3, PRDM5, PRKG1, PYCR1, RIN2, SKI, SLC26A2, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, SPARC, TALDO1, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, UPF3B, ZNF469
4151	小児 遺伝学	Invitae RASopathiesおよびヌーナンスペクトラム障害パネル Invitae RASopathies and Noonan Spectrum Disorders Panel	A2ML1, ACTB, ACTG1, BRAF, CBL, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NRAS, NSUN2, PPP1CB, PTPN11, RAF1, RASA1, RASA2, RIT1, RRAS, RRAS2, SHOC2, SOS1, SOS2, SPRED1, YWHAZ
5142	小児 遺伝学	InvitaeMACおよび前眼部形成異常パネル Invitae Microphthalmia, Anophthalmia, Coloboma (MAC) and Anterior Segment Dysgenesis Panel	ABCB6, ADAMTS18, ALDH1A3, ALX1, ASPH, BCOR, BMP4, BMP7, C12ORF57, CDON, CHD7, COL4A1, CPAMD8, CRYAA, CRYBA4, CYP1B1, DCDC1, ELP4, ERCC2, ERCC5, ERCC6, FAT1, FOXC1, FOXE3, FOXL2, FRAS1, FREM1, FREM2, FZD5, GDF6, GJA1, GRIP1, HCCS, HESX1, HMGB3, HMX1, ITPR1, KERA, MAB21L2, MFRP, MIR204, MITF, NAA10, NDP, OCRL, OTX2, PAX2, PAX6, PITX2, PITX3, PORCN, PRDM5, PRSS56, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, RBP4, RERE, SALL2, SHH, SIX3, SIX6, SLC38A8, SMCHD1, SMOC1, SOX2, STRA6, TBC1D20, TENM3, TFAP2A, UBE3B, VAX1, VSX1, VSX2, WNT2B, YAP1, ZDBF2, ZIC2
55013	小児 遺伝学	Invitaeスティックラー症候群パネル Invitae Stickler Syndrome Panel	COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, LOXL3, LRP2, VCAN
4612	小児 遺伝学	Invitae アントレー・ビクスラー症候群検査 Invitae Antley-Bixler syndrome Test	POR

Test Code	項目名
FVT0001	Family Variant Test

*** Family Variant Testの検査対象期間は結果報告後90日間です。**
*** Family Variant Testは発端者の陽性遺伝子に関する検査が可能です。**

Invitaeでは、カスタマイズに対応した フレキシビリティの高い遺伝学的パネル検査を設計し 疾患領域ごとに提供しています。

★ 疾患領域ごとに1サンプルあたりのシークエンス解析を単一価格で行っています。これにより、キュレーション済みシークエンスパネルを選択したり、複数のシークエンスパネルを組み合わせたり、またはサンプルごとにシークエンスパネルをカスタマイズ（再依頼*を含む）する場合も解析対象の遺伝子が1つの疾患領域に含まれる限り、表示価格のままで解析することができます。

★ シークエンスパネルには、特に断りのない限り、1サンプルの遺伝子解析、エクソンレベルの欠損/複数解析一式が含まれます。

★ 疾患領域が異なる2つの遺伝学的検査をご依頼いただいた場合は、遺伝学的検査が2種類有効になり、サンプル費用も2アッセイ分必要となります。また、異なる2つの専門チームにより2つのレポートを作成することになります。

★ 本一覧表に掲載された全キュレーション済みシークエンスパネルは、予備的証拠及びアドオンを含め、全てそれぞれの疾患領域に帰属します。

* 再依頼とは、初回のシークエンス解析と同じ疾患領域の遺伝学的検査について、追加料金の支払いやサンプルの新規提供なしで追加のオーダーができるサービスです（レポート受領から90日以内）。本サービスにより、適切な遺伝学的検査を適切なペースでフレキシブルにオーダーすることが可能となります。

*本検査内容は新しい臨床知見、ガイドライン、専門家の意見を検討した上でInvitae社の方針で変更になる可能性があります。

詳細については、下記の連絡先までお問い合わせ下さい。

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