U.O.C. GENETICA MEDICA

ELENCO DELLE PRESTAZIONI DI GENETICA MOLECOLARE A SCOPO DIAGNOSTICO

area diagnostica	patologia /disease	geni testati	metodo di analisi
	carcinoma midollare della tiroide	RET	pannello NGS regione genomica locus RET
enteropatie	Hirschsprung Disease (HSCR)		
	Pseudo-ostruzione intestinale (CIPO)	ACTG2	Sanger Seq
		MYH11	pannello ampliconi NGS
reumatologica	sindromi auto-infiammatorie (SAID) (pannello base)	IL1RN, LPIN2, MDFIC, MEFV, MVK, NLRP12, NLRP3, NOD2, PSMB8, PSTPIP1, TNFRSF1A	pannello NGS "FP11"
	sindromi auto-infiammatorie (SAID) (pannello avanzato)	ACP5, ADAR1, AP1S3, C1NH, CARD14, CECR1, Dnase1, DNase1L3, DNase2, IFIH1, IL10, IL10RA, IL10RB, IL1RN, IL36RN, ISG15, LPIN2, MEFV, MVK, NLRC4, NLRP12, NLRP3, NLRP7, NOD2, PLCG2, PSMA3, PSMB4, PSMB8, PSMB9, PSTPIP1, RBCK1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SLC29A3, SH3BP2, TMEM173, TNFRSF1A, TNFRSF11A, TREX-1	pannello NGS "FP41"
emato-immuno-reumatologica	sindromi emato-immuno-reumatologiche	FAS, FASL, CASP8, CASP10, KRAS, NRAS, MAGT1, FADD, ITK, CTLA4, LRBA, PIK3CD, PIK3R1, CARD11, TNFRSF13B, TNFRSF13C, CD19, CD20, CD40, CD40L, PRKCD, DKC1, CTC1, RPL5, NOP10, RTEL1, RPL11, TERT, TINF2, RPL35A, RPS10, RPS19, RPS24, RPS26, SBDS, WRAP53, RPS7, RPS17, RPL26, NHP2, ELA2, GFI1, HAX1, G6PC, VPS45, CSF3R, JAGN1, VPS13B, AP3B1, C16orf57, CXCR4, LAMTOR2, LYST, RAB27A, RAC2, SLC37A4, TAZ, WAS	pannello NGS (58 geni)
stroke	sindromi con stroke quale manifestazione clinica comune	ABCC6, ACTA2, ATP7A, CBS, CECR1, COL4A1, ELN, GLA, HTRA1, JAG1, NF1, NOTCH3, PCNT, SAMHD1, SLC2A10	pannello NGS (15 geni)
		ABCA1, ABCC6, ABCG2, ACE, ACTA2, ACVRL1, ADAMTS17, ADAR, ANK1, APOA1, APOA2, APOA4, APOA5, APOB, APOE, APP, ARHGAP10, ATP1A2, ATP1A3, ATP7A, BHMT, BRCC3, CACNA1A, CBS, CCM2, CECR1, COL1A1, COL1A2, COL3A1, COL4A1, COL4A2, COL5A1, COL5A2, CST3, ELN, ENG, ENPP1, F13A1, F13B, F2, F5, F7, F8, F9, FBLN5, FBN1, FGA, FGB, FGG, FLNA, GGCX,	pannello NGS (127 geni)

		GLA, GNAQ, GP1BA, GP6, GUCY1A3, HBA1, HBA2, HBB, HTRA1, IFIH1, ITGA2, ITGB1, ITGB2, ITGB3, ITM2B, JAG1, KNG1, KRIT1, LPL, MCFD2, MMADHC, MMP3, MTHFR, MTR, MTRR, NF1, NOS3, NOTCH3, PDCD10, PDE4D, PDGFRB, PKD1, PKD2, PLAT, PLAU, PLAUR, PLG, PLOD1, PMF1, PON1, PROC, PROCR, PROS1, RASA1, RNASEH2A, RNASEH2B, RNASEH2C, RNF213, SAMHD1, SCN1A, SERPINA5, SERPINC1, SERPIND1, SERPINE1, SKI, SLC1A2, SLC25A44, SLC2A10, SLC44A2, SLC4A1, SMAD3, SMAD4, SPTA1, SPTB, STAT3, STXBP5, TGFB1, TGFB2, TGFBR1, TGFBR2, THBD, TIMP2, TREX1, TSPAN15, VEGFA, VWF	
	stroke post-evento infettivo (varicella)	ABCA1, ACE, ADAMTS17, ADAR, APOA1, APOA2, APOA4, APOA5, APOB, BHMT, CBS, CECR1, F13A1, F13B, F2, F5, F7, F8, F9, FGA, FGB, FGG, GP1BA, GP6, ITGA2, ITGB3, KNG1, LPL, MCFD2, MMADHC, MTHFR, MTR, MTRR, NOS3, PDGFRB, PLAT, PLAU, PLAUR, PLG, POLR3A, PON1, PROC, PROS1, SAMHD1, SERPINC1, SERPIND1, SERPINE1, STAT3, THBD, VWF. POLR3C*, POLR3F* * sequenziamento Sanger	pannello NGS (50 geni) e Sanger seq
Malattie rare	ipoventilazione centrale congenita (CCHS)	PHOX2B	Sanger Seq
	Alexander Diasease	GFAP	Sanger Seq
Malattie rare/Quadri sindromici congeniti	Nail-Patella Syndrome (NPS, 161200)	*LMX1B*	Sanger Seq
	Fibrodysplasia Ossificans Progressiva (FOP, 135100)	*ACVR1*	Sanger Seq
	EEC and related disorders	*TP63*	Sanger Seq
	Oloprosencefalia	SHH	Sanger Seq
Malattie ad eziologia sconosciuta	Malattie complesse congenite o ad esordio infantile	Analisi dell'esoma che comprende tutte le regioni codificanti, le regioni di splicing , le regioni non tradotte (UTR) al 5' ed al 3' dei geni codificanti ed alcune regioni introniche di particolare interesse clinico.	NGS
Farmaco-genetica	Reazioni avverse al trattamento con farmaci a base di Tiopurine	TPMT	Sanger Seq