U.O.S.D. LABORATORIO DI GENETICA E GENOMICA DELLE MALATTIE RARE

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
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, 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	pannello NGS (127 geni)

	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 non diagnosticate	Whole Exome Sequencing (WES) e Whole Genome Sequencing (WGS)	NGS