Servizio Sanitario della Toscana

CATALOGO PRESTAZIONI

U.O.C. GENETICA MEDICA

AOU SENESE

PRESTAZIONI GENETICA MOLECOLARE

Prestazioni che vengono tariffate all'arrivo del campione

CAMPIONE BIOLOGICO	METODICA	CODICE PRESTAZIONE AOUS	Tempi di Risposta	CODICE PRESTAZIONE regionale		Tariffa
DNA/RNA	Conservazione DNA/RNA	K1598	3gg	91.36.1		41
Sangue o altro tessuto	estrazione DNA conservazione DNA	K1602 K1598	3gg	91.36.5 91.36.1		46 41
Villi coriali o altro tessuto	Analisi di Polimorfismi STR con PCR e elettroforesi capillare	K1565	7 gg	91.29.3		57
Carcinoma gastrico	CDH1	NGS	K1570x10	2 mesi	91.30.3x10	156x10
Carcinoma mammella e ovaio	BRCA1, BRCA2	NGS	K1570x20	3 mesi	91.30.3x20	156x20
Carcinoma mammella e ovaio in URGENZA	BRCA1, BRCA2	NGS	K1570x20	21 giorni	91.30.3x20	156x20
Carcinoma mammella e ovaio	BRCA1(del/dup) BRCA2 (del/dup)	MLPA	K1609Qx4	1 mese	91.38.2x4	121x4
Carcinoma midollare della Tiroide (FMTC)	RET	NGS	K1570lx20	3 mesi	91.30.3x20	156x20
Lynch, Sindrome di	MLH1 MSH2 MSH6 PMS2	NGS	K1570x8	4 mesi	91.30.3x8	156x8NGS

	EPCAM (3'UTR)					
Lynch, Sindrome di	MLH1, MSH2 (del/dup) PMS2 (del/dup) MSH6, EPCAM (del/dup)	MLPA	K1609Qx6	1 mese	91.38.2x6	121x6
Pannello tumori	BRCA1, BRCA2, RB1, TP53, APC, CDH1, MLH1, MSH2, MSH6, PMS2, PTEN, CDKN2A, ATM, CHEK2, BRIP1, PALB2, BAP1, DICER1, BMPR1A, SMAD4, STK11, ENG, POLD1, POLE, CDK4, VHL, FH, FLCN, MUTYH, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, MEN1, RET, NF1, NF2, SUFU, PTCH1, PRKAR1A, TSC1, TSC2, RAD51C, RAD51D	NGS	K1570x40	4 mesi	91.30.3x40	156x40
Poliposi familiare adenomatosa, forma attenuata inclusa	APC MUTYH	NGS	K1570x6	2 mesi	91.30.3x6	156x6
Retinoblastoma	RB1	NGS	K1570x10	3 mesi	91.30.3x10	156x10
Retinoblastoma	RB1 (del/dup)	MLPA	K1609Qx2	1 mese	91.38.2	121x2
Test mammella secondo livello/tumori multipli	PTEN, p53	NGS	K1570x8	2 mesi	91.30.3x8	156x5
Tumori stromali gastro-intestinali (GIST)	KIT, PDGRFA, SDHA, SDHB, SDHC	NGS	k1570x20	3 mesi	91.30.3x20	156x20
Test su DNA circolante per tumore del polmone	ALK, BRAF, MET, ROS1, EGFR, ERBB2, KRAS, TP53, PIK3CA, NRAS, MAP2K1, RET	NGS	K1570x7	7 giorni	91.30.3x7	156x7
Test su DNA circolante per tumore della	EGFR, ERBB2, KRAS, TP53, PIK3CA, AKT1, FBXW7, ERBB3, ESR1, F3B1, CCND1,	NGS	K1570x10	7 giorni	91.30.3x10	156x10

mammella	FGFR1					
Test su DNA circolante per tumore del colon	BRAF, EGFR, ERBB2, KRAS, TP53, PIK3CA, NRAS, MAP2K1, AKT1, FBXW7, CTNNB1, GNAS, SMAD4, APC	NGS	K1570x10	7 giorni	91.30.3x10	156x10
Test su DNA circolante per tutti i tipi di tumore	ALK, BRAF, MET, ROS1, EGFR, ERBB2, KRAS, TP53, PIK3CA, NRAS, MAP2K1, RET, AKT1, FBXW7, ERBB3, ESR1, CCND1, FGFR1, CTNNB1, GNAS, SMAD4, APC, AR, ARAF, CHEK2, FLT3, DDR2, FGFR2, FGFR3, FGFR4, GNA11, GNAQ, HRAS, IDH1, IDH2, KIT, MAP2K2, MTOR, NTRK1, NTRK3, PDGFRA, RAF1, SF3B1, CDK4, SMO, PTEN, CCND2, CCND3, CDK6, MYC, ERG, ETV1	NGS	K1570x10	7 giorni	91.30.3x10	156x10
CFTR, analisi di primo livello	CFTR	NGS	K1570x8	2 mesi	91.30.3x8	156x8
CFTR, analisi di secondo e terzo livello	CFTR	NGS	K1570x16	2 mesi	91.30.3x16	156x16
Microdelezioni del braccio lungo del cromosoma Y	Cromosoma Y	PCR ed Elettroforesi Capillare	K1565Lx3	3 mesi	91.29.3x3	57x3

Prestazioni che vengono tariffate al momento della stesura del referto

PATOLOGIA(E)	GENE(I)	METODICA	CODICE PRESTAZIONE AOUS	Tempi Risposta	CODICE PRESTAZIONE Regionale	Tariffa
Alcaptonuria	HGD	NGS	k1570	1 mese	91.30.3x2	156x2
Alport, Sindrome di	COL4A5	NGS	K1570x40	5 mesi	91.30.3x40	156x40
	COL4A4					
	COL4A3					
Alport, X-legata sindrome di	COL4A5 (del/dup)	MLPA	K1609Qx2	1 mese	91.38.2x2	121x2

Alport autosomico, sindrome di	COL4A4 (del/dup) COL4A3 (del/dup)	MLPA	K1609Qx4	1 mese	91.38.2x4	121x4
Aneurismi ereditari	MYH11, ACTA2, MYLK, SLC2A10, COL3A1, SMAD3, TGFBR1, TGFBR2, FBN1, COL4A1	NGS	K1570x40	5 mesi	91.30.3x40	156x40
Sindrome di Angelman	Regione 15q 11-13	MLPA metilazione inclusa	K1609Q	1 mese	91.38.2	121
Anomalie cromosomiche criptiche "whole genome"	Tutti i cromosomi	Array-CGH	K1563x2	4 mesi	91.29.1x2	127x2
			K1571x2		91.30.4x2	116x2
			K1601		91.36.4	43
			K1603		91.37.1	82
Anomalie cromosomiche criptiche su regioni specifiche	Cromosomi (specifici segmenti)	MLPA	K1609Q	10 giorni	91.38.2	121
Anomalie cromosomiche di numero	Cromosomi 13, 18, 21, X, Y	QF-PCR	K1570L	4 giorni	91.30.3	156
Anomalie cromosomiche di numero su materiale abortivo	Cromosomi 13, 15, 16, 18, 21, 22, X, Y	QF-PCR	K1570Lx2	3 mesi	91.30.3x2	156x2
Aritmie ereditarie QT-lungo, QT-corto, Sindrome di Brugada inclusi	KCNQ1, KCNH2, SCN5A, ANK2, KCNE1, KCNE2, KCNJ2, CACNA1C, CAV3, SCN4B, AKAP9, SNTA1, KCNJ5, GPD1L, SCN1B, KCNE3, SCN3B, HCN4	NGS	K1570x25	4 mesi	91.30.3x25	156x25
Cheratocono	VSX1, HGF, CRB1, DOCK9, AIPL1, SOD1, RAB3GAP1	NGS	K1570x 5	3 mesi	91.30.3x5	156x5
Cohen, Sindrome di	COH1	NGS	K 1570x20	3 mesi	91.30.3x20	156x20
Cohen, Sindrome di	COH1(del/dup)	MLPA	K1609Qx2	1 mese	91.38.2 x2	121x2
Deficit intellettivo e microcefalie	ABCD1, ACSL4, AFF2, AGTR2, AP1S2, ARHGEF6, ARHGEF9, ARX, ASPM, ATP6AP2, ATP7A, ATRX, BCOR, BRWD3, CASK, CC2D1, CCDC22, CDH15, CDK16, CDK5RAP2, CDKL5, CENPJ, CEP135, CEP152, CHD8, CLCN4, CLIC2, CNKSR2, CNTNAP2, CRBN, CSTF2, CTNNB1, CUL4B, DCX, DKC1, DLG3, DOCK8, DYNC1H1, EBP, EIF2S3, ELP2, FANCB, FGD1, FLNA, FMR1, FOXG1, FOXP1, FRMPD4, FTSJ1, GATAD2B, GDI1, GK, GPC3, GRIA3, GRIK2, GRIN2B, HCCS, HCFC1,	NGS	K1570x30	5 mesi	91.30.3x30	156x30

	HDAC6, HDAC8, HPRT1, HSD17B10, HUWE1, IDS, IGBP1, IKBKG, IL1RAPL1, IQSEC2, KATNAL2, KDM5C, KIAA2022, KIRREL3, KLF8, KLHL15, L1CAM, LAMP2, LAS1L, MAGT1, MAN1B1, MAOA, MAP2K1, MBD5, MBTPS2, MCPH1, MECP2, MED12, MED13L, MED23, MEF2C, MID1, MTM1, NAA10, NDP, NDUFA1, NHS, NLGN3, NLGN4X, NRXN1, NSDHL, NXF5, OCRL, OFD1, OPHN1, OTC, PAK3, PCDH19, PDHA1, PGK1, PHF6, PHF8, PLP1, PORCN, PQBP1, PRKRA, PRPS1, PRSS12, RAB39B, RAB40AL, RBM10, RLIM, RPL10, RPS6KA3, SATB2, SCN2A, SCN8A, SETBP1, SHANK2, SHANK3, SHROOM4, SLC16A2, SLC2A1, SLC6A8, SLC9A6, SMC1A, SMS, SOBP, SOX3, SRPX2, ST3GAL3, STIL, STXBP1, SYN1, SYNGAP1, TAF1, TAF7L, TCF4, TECR, THOC2, TIMM8A, TRAPPC9, TSPAN7, TUSC3, UBE2A, UPF3B, USP27X, WDR62, ZDHHC15, ZDHHC9, ZNF41, ZNF526, ZNF674, ZNF81					
Displasia aritmogena ventricolo destro	PKP2, DSP, JUP, TMEM43, DSC2, DSG2	NGS	K1570x10	3 mesi	91.30.3x10	156x10
Disturbi del linguaggio/disprassia verbale con o senza disabilità intellettiva e autismo	AP1S2, ARX, ASPM, ATRX, BRAF, CASK, CDKL5, CNTNAP2, CHD7, CREBBP, DCX, DHCR7, DMD, EHMT1, ERCC6, ERCC8, FGD1, FMR1, FOXG1, FOXP1, HDAC8, HOXA1, HPRT1, HRAS, KDM5C, KRAS, L1CAM, MAP2K2, MBD5, MECP2, MED12, MEF2C, MID1, MKKS, NF1, NIPBL, NRAS, NRXN1, NSD1, OPHN1, PCDH19, PQBP1, PTCH1, PTEN, PTPN11, RAB39B, RAD21, RAF1, RAI1, RELN, RPGRIP1L, RPS6KA3, SCN1A, SHANK3, SHOC2, SLC2A1, SLC9A6, SMC3, SOS1, SPRED1, TCF4, TSC1, TSC2, TUBA1A, UBE3A, VPS13B, ZEB2 ABCC9,ABCD1, ABCD4, ABHD5, ACAD9, ACOX1, ACSF3, ACSL4, ACTB, ACTG1, ACVR1, ACY1, ADAR, ADCK3, ADK, ADSL, AFF2, AGA, AGPAT2, AGTR2, AHCY, AHI1, AIFM1, AIMP1, AK1, AKT3, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALG1, ALX1, ALX4, AMT, ANKH, ANKRD11, ANO10, AP3B1, AP4B1, AP4E1, AP4M1, AP4S1, APTX, ARFGEF2, ARG1, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARL13B, ARL6, ASL, ASPA, ASXL1, ATIC, ATP1A2, ATP2A2, ATP6AP2, ATP6V0A2, ATP7A, ATR, AUH, B3GALTL, B4GALT1, B4GALT7, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCKDHA, BCKDHB, BCOR, BCS1L, BLM, BRWD3, BSCL2, BTD, BUB1B, C12orf57, C5ORF42, CA2, CA8, CACNA1C, CACNG2, CBL, CBS, CC2D1A, CC2D2A, CCBE1, CCDC78, CDH15, CDK5RAP2, CDON, CENPJ, CEP135, CEP152, CEP290, CEP41, CHD2, CLCNKB, CLIC2, CLN3, CLN5, CLN6, CLN8, COG1, COG7, COG8, COL4A1, COL4A2, COLEC11, COQ2, COX15, CPS1, CRBN, CTDP1, CTNNB1, CTSA, CTSD, CUL4B, CYB5R3, D2HGDH, DARS2, DBT, DCAF17, DDHD2, DHCR24, DHFR, DHTKD1,	NGS	K1570x40	6 mesi	91.30.3x40	156x40

	DIP2B, DKC1, DLD, DLG3, DMPK, DNAJC19, DNMT3B, DOCK8, DPAGT1, DPM1, DPYD, DST, DYM, DYNC1H1, DYRK1A, EFTUD2, EIF2AK3, ELOVL4, EMX2, EPB41L1, ERCC2, ERCC3, ERCC5, ERLIN2, ESCO2, ETHE1, FAM126A, FBN1, FGFR1, FGFR2, FGFR3, FH, FKRP, FKTN, FLNA, FRAS1, FTO, FTSJ1, FUCA1, GAD1, GALE, GALT, GAMT, GATAD2B, GATM, GCH1, GCSH, GDI1, GFAP, GJB1, GJC2, GK, GLB1, GLDC, GLI2, GLI3, GM2A, GNAS, GNPAT, GNS, GPC3, GPHN, GPR56, GRIA3, GRIK2, GRIN1, GRIN2A, GRIN2B, GRM1, GSS, GTF2H5, GUSB, HAX1, HCCS, HCFC1, HDAC4, HDAC6, HESX1, HEXA, HEXB, HLCS, HPD, HSD17B10, HSPD1, HUWE1, IDS, IDUA, IER3IP1, IGBP1, IGF1, IKBKG, IL1RAPL1, INPP5E, IQSEC2, ISPD, JAM3, KANSL1, KAT6B, KCNJ10, KCNJ11, KCNK9, KCNQ2, KCNT1, KCTD7, KDM6A, KIAA1279, KIF11, KIF1A, KIF7, KIRREL3, L2HGDH, LAMA2, LAMC3, LAMP2, LARGE, LARP7, LIG4, LRP2, LRPPRC, MAGT1, MAN1B1, MAN2B1, MANBA, MAOA, MAP2K1, MAT1A, MCCC1, MCCC2, MCOLN1, MCPH1, MED17, MED23, MGAT2, MLYCD, MMAA, MMACHC, MMADHC, MOCS1, MOCS2, MPDU1, MPLKIP, MRPS22, MTR, MTRR, MUT, MVK, MYCN, MYO5A, NAA10, NAGA, NAGLU, NALCN, NDE1, NDP, NDUFA1, NDUFA11, NDUFA12, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NEU1, NKX2-1, NLRP3, NPHP1, NSDHL, NSUN2, NTRK1, OCLN, OCRL, OFD1, ORC1, OTC, PACS1, PAH, PAK3, PANK2, PAX6, PAX8, PC, PCNT, PDHA1, PDSS1, PDSS2, PEPD, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP2, PGK1, PHF6, PHF8, PHGDH, PIGN, PIGO, PIGV, PIK3R2, PLA2G6, PLCB1, PLP1, PMM2, PNKP, PNP, POC1A, POLR3A, POLR3B, POMGNT1, POMT1, POMT2, PORCN, PPOX, PPT1, , PRODH, PRPS1, PRSS12, PSAP, PUS1, PVRL1, PYCR1, RAB18, RAB27A, RAB3GAP1, RAB3GAP2, RAB40AL, RARS2, RBM10, RBM28, RFT1, RIT1, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, ROGDI, SALL1, SATB2, SC5D, SCN2A, SCN8A, SCO2, SDHA, SERAC1, SETBP1, SHH, SHROOM4, SIL1, SIX3, SKI, SLC12A6, SLC16A2, SLC17A5, SLC25A15, SLC25A22, SLC33A1, SLC35C1, SLC4A4, SLC6A8, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMOC1, SMPD1, SMS, SNAP29, SOBP, SOX10, SOX3, SPG11, SPTAN1, SRCAP, SRD5A3, SRPX2, ST3GAL3, STIL, STRA6, STXBP1, SUOX, SURF1, SYN1, SYNGAP1, SYP, SYT14, TAT, TBC1D24, TBCE, TECR, TGFBR1, TGFBR2, THRB, TIMM8A, TMCO1, TMEM165, TMEM231, TMEM237, TMEM67, TPP1, TRAPPC9, TREX1, TSPAN7, TTC8, TUBA8, TUBB2B, TUSC3, UBE2A, UBR1, UPB1, UPF3B, VLDLRVRK1, WDR45, WDR62, XPA, XPNPEP3, ZBTB16, ZDHHC9, ZIC2,					

	ZNF41, ZNF592, ZNF674, ZNF711, ZNF81					
Encefalopatie mitocondriali	AARS2 C10orf2 DGUOK EARS2 ETHE1 MPV17 MTFMT MTO1 NDUFS2 NDUFS4 PDHA1 POLG RMND1 RRM2B SUCLA2 SUCLG1 SURF1 TK2 TMEM70 TRMU AGK AIFM1 ATP5E ATPAF2 AUH BCS1L BOLA3 C10orf2 COX14 COX15 COX4I2 COX6B1 DARS2 DLAT DLD DNAJC19 EARS2 EIF2AK3 ETFA ETFB ETFDH ETHE1 FARS2 FASTKD2 FOXRED1 GFAP GFER GFM1 IBA57 LRPPRC MARS2 MRPS16 MRPS22 MTFMT MTO1 MTPAP NDUFA12 NDUFA1 NDUFA2 NDUFA9 NDUFAF2 NDUFAF3 NDUFAF4 NDUFAF5 NDUFAF6 NDUFB3 NDUFB9 NDUFS1 NDUFS2 NDUFS3 NDUFS4 NDUFS6 NDUFS7 NDUFS8 NDUFV1 NDUFV2 NFU1 NUBPL OPA1 PANK2 PC PDHA1 PDHB PDHX PDP1 PDSS1 PDSS2 PNPT1 PUS1 RARS2 RMND1 RRM2B SARS2 SCL19A2 SCO1 SCO2 SDHA SDHAF1 SDHAF2 SDHB SDHC SDHD SERAC1 SLC19A3 SLC25A12 SLC25A19 SLC25A20 SLC25A3 SLC25A4 SLC33A1 SLC6A8 SPG7 SUCLA2 SUCLG1 SURF1 TACO1 TAZ TIMM8A TMEM70 TPK1 TRMU TSFM TTC19 TUFM TYMP UQCRB UQCRC2 UQCRQ WFS1 YARS2	NGS	K1570x40	6 mesi	91.30.3x40	156x40
Febbre mediterranea familiare	MEFV	NGS	K1570x 5	2 mesi	91.30.3x 5	152 x 5
Fibrosi Polmonare	TERC1 TERT SFTPC	NGS	K1570x10	3 mesi	91.30.3x10	156x10
Floating- Harbor, Sindrome	SRCAP	NGS	K1570x8	4 mesi	91.30.3x8	15dx8
Glaucoma angolo aperto	MYOC OPTN	NGS	K1570x10	3 mesi	91.30.3x10	156x10
Huntington, Corea di	HD	PCR ed Elettroforesi capillare	K1565Lx2	2 mesi	91.29.3x2	57x2
Huntington, Corea di diagnosi presintomatica	HD	PCR ed Elettroforesi capillare	K1565Lx4	3 mesi	91.29.3x4	57x4
Ittiosi X-legata	STS	PCR	K1565	10 gg	91.29.3	57
Kallmann, Sindrome di	KAL1	PCR	K1565	10 gioni	91.29.3	57
Lesch-Nyhan	HPRT1	NGS	K1570x9	3 mesi	91.30.3x9	156x9
Leucoencefalopatie	ABCD1, AIMP1, ARSA, ASPA, ATP13A2, C19ORF12, CP, CSF1R, CYP27A1, DARS2, DCAF17, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, FA2H, FAM126A (also known as DRCTNNB1A or HCC), FTL, GALC, GFAP, GJC2, HEPACAM, HSD17B4, HSPD1, LMNB1, MLC1, NDUFV1, PANK2, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2,	NGS	K1570x8	4 mesi	91.30.3x8	156x8

	PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PLA2G6, PLP1, POLR3A, POLR3B, PSAP, RNASET2, SCP2, SLC16A2, SOX10, TREM2, TREX1, TYROBP					
Linfedema primario ereditario	GJC2, KIF11, FOXC2, CCBE1, MET, GATA2, FLT4, HGF, SOX18, VEGFC, GJA1, ITGA9	NGS	K1570x20	4 mesi	91.30.3x20	156x20
Malattia di Parkinson	ADH1C, ATP13A2, ATXN2, FBXO7, GBA, GIGYF2, GLUD2, HTRA2, LRRK2, MAPT, NR4A2, PARK2, PINK1, PLA2G6, SNCA, SNCAIP, TBP,UCHL1, VPS35, DJ1, SLC6A3, DNAJC6, SYNJ1, TAF1, EIF4G1	NGS	K1570x20	3 mesi	91.30.3x20	156x20
Malattie Autoinfiammatorie	MEFV, TNFRSF1A,, NLRP3, NLRP12,, MVK, NOD2, IL36RN, PSTPIP1, LPIN2, ADA2, TNFAIP3, NLRC4, IL1RN, OTULIN, PLCG2, RBCK1, SH3BP2, SLC29A3, CARD14, AP1S3, TMEM173, PSMA3, PSMB4, PSMB8, PSMB9	NGS	K1570x40	6 mesi	91.30.3x40	15dx40
Malformazioni Arterovenose (MAV)	GNA11, GNAQ, AKT1, KRAS, HRAS, IDH1, PIK3CA, TEK, PTEN, RASA1, TGFBR2, TGFBR1, TGFB1, TGFB2, SMAD3, SMAD4, GLMN	NGS	K1570x40	6 mesi	91.30.3x40	15dx40
Mielodisplasie	CALR, JAK2, MPL	NGS	K1570x 5	3 mesi	91.30.3x5	156x5
Neuropatie ereditarie periferiche	AARS, ABHD12, AIFM1, ATL1, ATP7A, BSCL2, C10orf2, COX6A1, CTDP1, DCTN1, DHH, DHTKD1, DNAJB2, EXOSC8, FAM134B, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GNB4, HINT1, HOXD10, HSPB1, HSPB8, IGHMBP2, IKBKAP, INF2, KARS, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MED25, MFN2, MPZ, MTMR2, NDRG1, NEFL, NGF, NTRK1, PHYH, PLEKHG5, PMP22, PRPS1, PRX, RAB7A, SBF2, SCN10A, SCN11A, SCN9A, SEPT9, SH3TC2, SLC12A6, SLC52A3, SLC5A7, SMN1, SOX10, SPTLC1, SPTLC2, SYT2, TDP1, TFG, TRPV4, TTR, VCP, WNK1, YARS, MARS, SIGMAR1, SLC25A46, SLC52A2, MME, MORC2, MPV17, SPG2, MYH14, SPTLC3, NAGLU, SURF1, NEFH, TRIM2, ITR, UBA1, PDK3, VAPB, PMP2, VRK1, WNK1, PRDM12, REEP1, SBF1, SCNIIA, SETX, AIFM1, FBLN5, ARHGEFIO, FBX038, ASAH1, ATL3, FLVCR1, BICD2, CIOORF2, C120RF65, GJB3, CCT5, CHCHDIO, HARS, CLTCL1, HEXB, CRDP1, HK1, DCAF8, HOXDIO, DCTN2, HSPB3, DGAT2, DGUOK, DNM2, DNMT1, KIFIA, DRP2, KIFIB, DYNCIH1, EGR2, LASIL, EXOSC3	NGS	K1570x20	4 mesi	91.30.3x20	156x20
Neuropatia con paralisi da pressione(HNPP)/Charcot-Marie-Tooth di tipo 1A (CMT1A), inclusa	PMP22	Real-Time	K1609	3 mesi	91.38.2	121
Nicolaides-Baraitser, /	SMARCA2 SMARCB1	NGS	K1570x30	4 mesi	91.30.3x30	156x30

Coffin-Siris, Sindrome di	SMARCE1 ARID1A ARID1B					
Oculofaringea, distrofia	PABPN1	PCR ed Elettroforesi capillare	K1565x2	2 mesi	91.29.3x 2	57x2
Oculofaringea, distrofia diagnosi presintomatica	PABPN1	PCR ed Elettroforesi capillare	K1565x3	2 mesi	91.29.3x 3	57x3
Pannello multigenico piccolo	Pochi geni	NGS	K1570x5	2 mesi	91.30.3x 5	156x5
Pannello multigenico grande	Molti geni	NGS	K1570x20	4 mesi	91.30.3x20	156x20
Prader-Willi, Sindrome di	Regione 15q11-13	MLPA (metilazione inclusa)	K1609	1 mese	91.38.2	121
Retinoschisi X-legata	XLRS1	NGS	K1570x6	2 mesi	91.30.3x6	156x6
Rett, sindrome di e varianti	MECP2 CDKL5 FOXG1 MEF2C	NGS	K1570x20	3 mesi	91.30.3x20	156x20
Rett classica e variante Zappella	MECP2(del)	MLPA	K1609lx2	1 mese	91.38.2x2	121x2
Rett variante a convulsioni precoci	CDKL5 (del)	MLPA	K1609lx2	1 mese	91.38.2x2	121x2
Rett variante congenita	FOXG1(del)	MLPA	K1609lx2	1 mese	91.38.2x2	121x2
Test di esclusione contaminazione materna	24 loci genomici	Analisi di polimorfismi STR	K1570L	4 giorni	91.30.3	156
UPD (disomia uniparentale)	UPD per singolo cromosoma (es 14 oppure 15 oppure 7)	Elettroforesi capillare	K1570x5	20 giorni	91.30.3x20	156x5