Department of Medical Genetics - Laboratory of Molecular Genetics, Institute of Mother and Child						
DIAGNOSTICS USING NEXT GENERATION SEQUENCING - NGS (includes DNA isolation)						
EXOME SEQUENCING with analysis of cosegregation of selected variants in the family using Sanger sequencing (up to 3 chosen variants)						
Lp.	**disease**	***gene/region***	**OMIM**	**analysis range**	**procedure**	**price (EUR)**
1	Exome sequencing in a single individual		-	Analysis of clinically significant genes according to clinical diagnosis (verification of selected variants using the Sanger method)	GEN-66	1000
2	Analysis of NGS results from an external center		-	Analysis of clinically significant genes for the selected clinical diagnosis	GEN-67	375
3	Reanalysis of the results		-	Additional analysis of results from the GEN-66 procedure	GEN-67	375
4	Exome sequencing in 2 symptomatic individuals		-	Exome analysis for the selected clinical diagnosis - sequencing of 2 symptomatic individuals (sibling, proband + symptomatic parent)	GEN-66O	1375
5	Exome sequencing - TRIO		-	Exome analysis according to clinical diagnosis - sequencing of the proband and parents	GEN-66T	2000
PANEL SEQUENCING						
1	Any gene panel	based on exome sequencing	-	Analysis of a selected gene panel based on exome sequencing	GEN-66K	880
INFERTILITY						
2	Male/female infertility	gene panel	-	Analysis of 20 genes (list available on request) related to infertility	GEN-66N	450
NEUROLOGY						
3	Intellectual disability	based on exome sequencing	-	Analysis of genes related to intellectual disability based on exome sequencing (list available on request)	GEN-66A	880
4	Autism spectrum disorders	based on exome sequencing	-	Analysis of genes related to autism spectrum disorders based on exome sequencing (list available on request)	GEN-66A	880
5	Leukodystrophies	based on exome sequencing	-	Analysis of genes related to leukodystrophies (list available on request)	GEN-66L	880
6	Microcephaly	based on exome sequencing	-	Analysis of genes associated with microcephaly (list available on request)	GEN-66L	880

7	Neural migration defects	based on exome sequencing	-	Analysis of genes associated with neuronal migration defects (list available on request)	GEN-66L	880
8	Epileptic encephalopathies	gene panel	-	Panel of 49 genes (list available on request)	GEN-66B	450
9			-	Panel of 49 genes (list available on request) + isolation and banking material from parents, possible examination of selected variants (confirmation of pathogenicity / carrier testing)	GEN-66BE	550
10	Neonatal panel	gene panel	-	Panel of 83 genes (list available on request), associated with infantile seizure disorders and early infantile epileptic encephalopathies (NBE v.1)	GEN-66H	500
11		gene panel	-	Infantile epilepsy (seizure disorders/epileptic encephalopathies) (NBE v.1) + securing material from parents and possible testing of selected variants (confirmation of pathogenicity / carrier test)	GEN-66HE	550
12	Dravet syndrome/Dravet-like syndrome	gene panel	-	*SCN1A + PCDH19, CHD2, HCN1, GABRB3*	GEN-66B	450
13	Angelman syndrome / Rett syndrome	gene panel	-	*UBE3A, MECP2, FOXG1, CDKL5*	GEN-66B	450
14	Dystonia/Parkinson disease	gene panel	-	Analysis of genes (list available on request) associated with Dystonia / Parkinson's disease based on exome sequencing	GEN-66C	450
15	Hypotonic infant panel	gene panel	-	Analysis of genes associated with hypotonia in children based on exome sequencing	GEN-66I	880
16	Hypotonic infant panel plus	gene panel + MLPA	-	Analysis of genes associated with hypotonia in children based on exome sequencing + MPLA test for SMA and PWS	GEN-66I+	1100
17	Tuberous sclerosis	gene panel	191100, 613254	*TSC1, TSC2*	GEN-66J	450
GENODERMATOSIS						
1	Epidermolysis bullosa and diseases with skin fragility/sensitivity	gene panel		33 genes (list available on request)	GEN-66F	450
2	Ichthyosis, Palmoplantar keratoderma	gene panel	-	69 genes (list available on request)	GEN-66F	450
3	Other genodermatosis	gene panel	-	*CARD14 , PORCN , SASH1 , SMARCAD1 , TAT*	GEN-66F	450
4	Hair structure abnormalities	gene panel	-	*APCDD1 , CDH3 , CLDN1 , DSG1 , DSG4 , ERCC2 , ERCC3 , GTF2H5 , HR , LIPH , LPAR6 , MBTPS2 , MPLKIP , RMRP , RPL21 , SNRPE , SPINK5 , ST14*	GEN-66F	450
5	Ectodermal dysplasia	gene panel	-	list of the genes available on request	GEN-66F	450

METABOLIC DISEASES						
1	Metabolic disease panel	gene panel	-	Maple syrup disease, propionic aciduria, methylmalonic aciduria/cobalamin disorders, methylmalonic aciduria with homocystinuria, phenylketonuria, hyperphenylalaninemia, BH4 deficiency, dopaminergic and serotonergic neurotransmission disorders (list available on request)	GEN-66M	450
2	Pancreatitis	gene panel	-	*CELA3B , CFTR , CPA1 , CTRC , CUZD1 , PNLIP , SPINK1 , TRPV6 , UBR1*	GEN-66M	450
3	Pancreatitis	gene panel	-	Analysis of 9 genes associated with pancreatitis *CELA3B , CFTR , CPA1 , CTRC , CUZD1 , PNLIP , PRRS1 , SPINK1 , TRPV6 , UBR1* + sequencing of exon 2 and 3 of the gene *PRSS1*	GEN-66M+	450
HEARING LOSS						
1	Non-syndromic hearing loss	gene panel	-	150 genes (list available on request)	GEN-66E	450
2	Alstrom syndrome	gene panel	203800	*ALMS1*	GEN-66E	450
3	Alport syndrome	gene panel	-	*COL4A3, COL4A4, COL4A5*	GEN-66E	450
4	Jervell i Lange-Nielsen syndrome	gene panel	-	*KCNE1, KCNQ1*	GEN-66E	450
5	Branchio-oto-renal (BOR) syndrome	gene panel	-	*EYA1, SIX1, SIX5*	GEN-66E	450
6	Perrault syndrome	gene panel	-	*TWNK, HARS2, HSD1784, LARS2, CLPP*	GEN-66E	450
7	Pendred syndrome	gene panel	-	*FOXI1, KCNJ10, SLC26A4*	GEN-66E	450
8	Stickler syndrome	gene panel	-	*COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3*	GEN-66E	450
9	Treacher-Collins syndrome	gene panel	-	*TCOF1, POLR1C, POLR1D*	GEN-66E	450
10	Usher syndrome	gene panel	-	*VLGR1, CDH23, CIB2, CLRN1, HARS, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A, WHRN*	GEN-66E	450
11	Waardenburg syndrome	gene panel	-	*EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10*	GEN-66E	450
12	Wolfram syndrome	gene panel	-	*WFS1, CISD2*	GEN-66E	450
CONGENITAL DISORDERS						
1	RASopathies	gene panel	-	20 genes (list available on request)	GEN-66D	400
2	Neurofibromatosis type I (von Recklinghausena disease) (NF1)	gene panel	-	*NF1 , SPRED1*	GEN-34C	400
3	Baraitser-Winter syndrome	gene panel	243310	*ACTB, ACTG1*	GEN-66D	400
4	Kabuki syndrome	gene panel	147920	*KDM6A, KMT2D*	GEN-66D	400
5	Rubinstein-Taybi syndrome	gene panel	180849	*CREBBP, EP300*	GEN-66D	400
6	Coffin-Lowry syndrome	gene panel	303600	*RPS6KA3*	GEN-66D	400
7	Coffin-Siris syndrome	gene panel	135900	*ARID1A, ARID1B, SMARCA4, SMARCB1, SMRCE1*	GEN-66D	400
8	Nicolaides-Baraister syndrome	gene panel	601358	*SMARCA2*	GEN-66D	400
9	Klippel-Feil syndrome	gene panel	118100	*GDF3, GDF6, MEOX1*	GEN-66D	400
10	Schwannomatosis	gene panel	615670	*LZTR1, SMARCB1*	GEN-66D	400
11	Floating-Harbor syndrome	gene panel	136140	*SRCAP*	GEN-66D	400

12	Craniosynostosis	gene panel	-	Gene panel based on Twigg, Wilkie, AJHG, 2015	GEN-66G	450
13	Marfan syndrome / Loeys-Dietz syndrome	gene panel	-	gene panel *(FBN1, TGFBR1, TGFBR2, TGFB2, TGFB3, SMAD3, FBN2)*	GEN-66G	450
14	Overgrowth panel	gene panel	-	Genes list available on request	GEN-66G	450
15	Short stature syndrome panel	gene panel	-	Genes list available on request	GEN-66G	450
16	Gorlin syndrome	gene panel	-	*PTCH1, PTCH2, SUFU*	GEN-66G	450
17	Facial dystosis panel	gene panel	-	Genes list available on request	GEN-66G	450
18	Osteogenesis imperfecta	gene panel	-	Genes list available on request	GEN-66G	450
19	Sceletal dysplasia	gene panel	-	Genes list available on request	GEN-66G	450
20	Auriculocondylar syndrome	gene panel	-	Genes list available on request	GEN-66G	450
21	Ehlers-Danlos syndrome	gene panel	-	Genes list available on request	GEN-66G	450
22	Congenital disorders	based on exome sequencing	-	Genes list available on request	GEN-66K	880
DIAGNOSTICS PERFORMED BY SANGER SEQUENCING, MLPA/MS-MLPA, TP-PCR, and FRAGMENT ANALYSIS METHODS						
1	DNA extraction from blood / other tissues	-	-	-	GEN-13	40
CFTR-RELATED DISEASES						
1	Cistic fibrosis (CF)	*CFTR*	219700	Testing for the carrier status of any one mutation in the CFTR gene	GEN-02A	90
2				Identification of the p.Phe508del mutation and the dele2.3(21kb) mutation and all other mutations (over 70) in exon 10	GEN-02B	90
3				MLPA test (P091)	GEN-02J	125
4				Analysis of all 27 exons of the CFTR gene	GEN-02F	620
5				Identification of approximately 700 mutations, including 16 mutations most frequently occurring in Poland	GEN-02H	170
6				Identification of over 500 rare mutations in the CFTR gene - analysis of exons 1-6b, 8, 9, 18 (supplement to the GEN2H procedure) - part 1	GEN-02G	225
7				Identification of over 500 rare mutations in the CFTR gene, analysis of exons 12, 14a-17a, 19, 22-24 (supplement to the GEN2H procedure) - part 2	GEN-02I	225
8				Badanie dwóch dowolnych mutacji w genie CFTR	GEN-02C	125
9	Pancreatitis	*CFTR, SPINK1, PRSS1*	167800	Gene analysis: CFTR (exon 10 + dele2.3 (21kb)), PRSS1 (exons 2 and 3), SPINK1 (exon 3)	GEN-03A	110
10				Gene analysis: CFTR (exons 4, 9-11 + dele2.3 (21kb)), PRSS1 (exons 2 and 3), SPINK1 (exon 3)	GEN-03B	225
11		*CTRC*		Analysis of exons 2,3,7	GEN-03C	150
12		*CPA1*	114850	Analysis of exons 7-10	GEN-03D	150
INFERTILITY						
1				Analysis of exon 10 of the CFTR gene	GEN-01A	75

2	Male infertility	*CFTR*	277180	Analysis of exons 4, 7, 9-11, including identification of the p.Phe508del and dele2.3 mutations (21kb)	GEN-01B	100
3		*AZF*	415000	Analysis of 6 loci on chromosome Y	GEN-01D	125
4				Analysis of 6 loci on chromosome Y (EXTENDED analysis in accordance with the EAA / EMQN guidelines)	GEN-01E	70
5		*AZF + CFTR*	-	Male infertility package: CFTR (Analysis of exons 4, 7, 9-11, including identification of the F508del and dele2.3 mutations (21kb) + basic AZF	GEN-01F	150
6	Fragile X premature ovarian failure (FXPOF)	*FMR1*	311360	Pre-screening test (possibility of obtaining an uninformative result)	GEN-04A	75
7				Analysis for premutation/mutation presence (TP-PCR)	GEN-04C	180
8	Recurrent pregnancy lost	*F2*	614390	Identification of c.*97G>A (20210G>A) mutation	GEN-56A	75
9		*F5*	614389	Identification of p.Arg534Gln (V Leiden, R506Q) mutation	GEN-56B	75
10		*F2. F5*	-	Identification of c.*97G>A (20210G>A) mutation in F2 gene and p.Arg534Gln (V Leiden, R506Q) mutation in F5 gene	GEN-56C	100
11		*MTHFR*	-	1298A>C and 677C>T polymorphism analysis	GEN-12	100
12		*F2, F5, MTHFR*	-	Analysis of genes: F2 [identification of c.*97G>A (20210G>A) mutation], F5 [identification of p.Arg534Gln (V Leiden, R506Q) mutation] and MTHFR (polymorphisms 677C>T and 1298A>C)	GEN-56D	150
13		*PAI1 (SERPINE1)*	-	Analysis of polimorphism 4G/5G in PAI1 (SERPINE1) gene	GEN-56E	80
14	Female reproductive failure	*F2, F5, MTHFR, PAI1 (SERPINE1), FMR1*	-	Analysis of genes: *F2* [identification of c.*97G>A (20210G>A) mutation], *F5* [identification of p.Arg534Gln (V Leiden, R506Q) mutation] and *MTHFR* (polymorphisms 677C>T and 1298A>C), *PAI1* (*SERPINE1*) (polymorphism 4G/5G), *FMR1* (Pre-screening test)	GEN-56F	150
15			-	Analysis of genes: *F2* [identification of c.*97G>A (20210G>A) mutation], *F5* [identification of p.Arg534Gln (V Leiden, R506Q) mutation] and *MTHFR* (polymorphisms 677C>T and 1298A>C), *PAI1* (*SERPINE1*) (polymorphism 4G/5G), *FMR1* [analysis for premutation/mutation (TP-PCR)]	GEN-56G	250
NEUROLOGY						
1	Fragile X syndrome (FXS)/Fragile X-associated primary ovarian failure (FXPOF)/Fragile X–associated tremor/ataxia syndrome (FXTAS)	*FMR1*	FraX 300624 FXTAS 300623 FXPOF	Pre-screening test with evaluation of the number of CGG repeats in the normal range	GEN-04A	80
2				Analysis for premutation/mutation (TP-PCR)	GEN-04C	180
3				MS-MLPA test (ME029) - males only	GEN-04D	150

4			FraX 300624	Coding sequence analysis (FraX recognition only)	GEN-04E	250
5	X-linked intellectual disability (XLID)	*ARX*	300419	Coding sequence analysis	GEN-21A	240
6				Identification of the most common mutations in exon 2	GEN-21B	90
7		*MECP2*	300260	MLPA test (P015, MECP2 duplication syndrome)	GEN-21C	125
8		*MRX*	nd	MLPA test (P106, 16 XLID genes)	GEN-21D	140
9	Friedreich ataxia	*FXN*	229300	Dynamic mutation identification	GEN-07A	120
10				Analysis of exons 1-5	GEN-07C	190
11				MLPA test (P316)	GEN-07B	125
12	Prader-Willi syndrome	15q11-q13	176270	MS-MLPA test (ME028)	GEN-08C	150
13				Microsatellite analysis (15q chromosome)	GEN-08B	220
14	Angelman syndrome	15q11-q13	105830	MS-MLPA test (ME028)	GEN-09C	150
15				Microsatellite analysis (15q chromosome)	GEN-09B	220
16		*UBE3A*		Analysis of exons 7-16	GEN-09G	240
17	Rett / Rett-like syndrome	*MECP2*	312750	Analysis of exons 2-4	GEN-29A	150
18				MLPA test (P015)	GEN-29B	125
19		*CDKL5*	300672	Coding sequence analysis	GEN-29C	375
20				MLPA test (P189)	GEN-29D	125
21		*FOXG1*	613454	Coding sequence analysis	GEN-29E	120
22	FG syndrome	*MED12*	305450	Analysis of exons 21-28, 37	GEN-62A	150
23	Macrocephaly/autism	*PTEN*	605309	Coding sequence analysis	GEN-72A	200
24	SLC2A-related disorders (GLUT1-defficiency syndromes)	*SLC2A1*	606777, 612126,	Coding sequence analysis	GEN-10A	225
25				MLPA test (P138)	GEN-10B	125
26	SCN1A-related disorders (Dravet syndrome, GEFS+)	*SCN1A*	607208, 604403	Coding sequence analysis	GEN-15A	620
				MLPA test (P137)	GEN-15B	125
27	Epilepsy, female restricted, with intellectual disability (PCDH19-related syndrome)	*PCDH19*	300088	Coding sequence analysis	GEN-48A	220
28				MLPA test (P330)	GEN-48B	125
29	Parkinson disease of early-onset (PARK2)	*PARK2*	600116	Coding sequence analysis	GEN-18A	320
30				MLPA test (P051, P052)	GEN-18B	200
31	Parkinson disease of early-onset (PARK6)	*PINK1*	605909	Coding sequence analysis	GEN-74A	200
32				MLPA test (P051, P052)	GEN-74B	200
33	Parkinson disease of early-onset (PARK7)	*DJ1*	606324	Coding sequence analysis	GEN-74C	175
34				MLPA test (P051, P052)	GEN-74B	200
35	Parkinson disease of late-onset (PARK8)	*LRRK2*	607060	Identification of the p.Gly2019Ser mutation	GEN-20A	80
36				Analysis of exons 30, 31, 34, 35, 41, 48 (identification of known pathogenic point mutations)	GEN-20B	170
37	Parkinson Disease of late-onset (PARK1 and 4)	*SNCA*	168601	Analysis of exons 2 and 3 (identification of known pathogenic point mutations)	GEN-22A	125
38				MLPA test (P051)	GEN-22B	125
39	Dystonia type 1 (DYT1)	*DYT1*	128100	Analysis of exon 5, identification of c.907_909delGAG mutation	GEN-17A	80

40		*DYT1, DYT6, DYT12, DYT16*	nd	MLPA test (P059)	GEN-17B	140
41	Dystonia with dyskinesia (DYT6)	*THAP1*	602629	Coding sequence analysis	GEN-25A	100
42		*DYT1, DYT6, DYT12, DYT16*	nd	MLPA test (P059)	GEN-25B	140
43	Dopa-responsive dystonia	*GCH1*	600225	Coding sequence analysis	GEN-63A	150
44				MLPA test (P099)	GEN-63B	140
45		*TH*	191290	Coding sequence analysis	GEN-64A	275
46				MLPA test (P099)	GEN-64B	140
47		*SPR*	182125	Coding sequence analysis	GEN-65A	125
48	Dystonia type 8 (DYT8)	*MR1 (PNKD)*	118800	Analysis of exon 1 (p.Ala7Val and p.Ala9Val mutations)	GEN-68A	75
49	Dystonia type 10 (DYT10)	*PRRT2*	614386	Identyfikacja mutacji c.649dupC	GEN-69A	75
50				Coding sequence analysis	GEN-69B	120
51	Myoclonic dystonia (DYT11)	*SGCE*	159900	Coding sequence analysis	GEN-49A	250
52				MLPA test (P099)	GEN-49B	140
53	Dystonia type 4 (DYT4)	*TUBB4A*	128101	Coding sequence analysis	GEN-80A	170
54	Pelizaues-Merzbacher disease (PMD)	*PLP1*	312080	Coding sequence analysis	GEN-28A	190
55			312920	MLPA test (P022)	GEN-28B	125
56	Lissencephaly, X-linked	*DCX*	300067	Coding sequence analysis	GEN-79A	175
57	GM3 synthase deficiency / infantile onset epilepsy syndrome	*ST3GAL5*	609056	Coding sequence analysis	GEN-82A	175
58	Peripheral neuropathy (Charcot-Marie-Tooth disease)	*PMP22, GJB1, MPZ*	nd	MLPA test (P405)	GEN-83A	125
HEARING LOSS						
1	Nonsyndromic Hearing Loss	*GJB2*	220290	Analysis of exon 2 and IVS1+1G>A mutation	GEN-05A	125
2				MLPA test (P163) (includes genes *GJB2* , *GJB6* , *GJB3* , *POU3F4* , *WFS1*)	GEN-05B	125
3		*GJB6*	604418	Analysis of coding exon of *GJB6*	GEN-05F	75
4				Analysis of the most common deletions in the *GJB6* gene: del(*GJB6* -D13S1830), del(*GJB6* -D13S1854) (PCR analysis)	GEN-05E	75
5	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct or Pendred syndrome	*SLC26A4*	605646	Analysis of selected exons (9-12 and 14)	GEN-05C	125
6				Analysis of exons: 2-8, 13, 15-21	GEN-05D	500
7	KID syndrome	*GJB2*	148210	Analysis of exon 2 of *GJB2* gene	GEN-37	80
8	Nonsyndromic Hearing Loss: DFNB16 and DFNB22	*STRC, OTOA*	-	MLPA test (P461)	GEN-05G	125
9	Deafness-infertility syndrome (DIS) syndrome	*STRC , CATSPER2*	611102	MLPA test (P461)	GEN-05G	125
NEUROMUSCULAR DISORDERS / FLOPPY CHILD syndrome						
1	Spinal muscular atrophy	*SMN1*	SMA-1 253300 SMA-2	Identification of exon 7 deletion of *SMN1* gene with analysis of *SMN1* and *SMN2* copy number - MLPA assay (P060 or P021)	GEN-06B	125
2			253550 SMA-3	Carrier testing (*SMN1* exon 7 deletion) - MLPA test (P060)	GEN-06B	125

3			253400	Coding sequence analysis	GEN-06C	170
4	Spinal muscular atrophy with respiratory distress (SMARD)	*IGHMBP2*	604320	Coding sequence analysis	GEN-55A	250
5				MLPA test (P058)	GEN-55B	125
6	Spinal muscular atrophy, X-linked	*UBA1*	301830	Analysis of exon 15	GEN-55C	75
7				Coding sequence analysis excluding exon 15	GEN-55D	400
8	Hereditary motor and sensory neuropathy	*TRPV4*	600175	Coding sequence analysis	GEN-55E	350
9	Spinal muscular atrophy, lower extremity-predominant, autosomal dominant	*BICD2*	615290	Coding sequence analysis	GEN-55F	225
10	Nemaline myopathy	*ACTA1*	161800	Coding sequence analysis	GEN-73A	125
11	Myotubular myopathy, X-linked	*MTM1*	310400	Coding sequence analysis	GEN-16	350
12	EMARDD syndrome	*MEGF10*	614399	Coding sequence analysis	GEN-55G	420
13	Emery-Dreyfuss muscular dystrophy	*LMNA/C*	150330	Coding sequence analysis	GEN-50A	275
14				MLPA test (P048)	GEN-50B	125
15		*EMD*	30384	Coding sequence analysis	GEN-50C	100
16		*FHL1*	300696	Coding sequence analysis	GEN-50D	140
RASOPATHIES						
1	Noonan syndrome	*PTPN11*	163950	Analysis of exons: 2-4, 7-9, 12, 13	GEN-19A	125
2	Cardio-facio-cutaneous syndrome (CFC)	*BRAF*	115150	Analysis of exons: 6, 11-17	GEN-26A	175
3	Costello syndrome (FCS)	*HRAS*	218040	Coding sequence analysis	GEN-33A	100
4	Neurofibromatosis type I (von Recklinghausena disease) (NF1)	*NF1*	162200	MLPA test (P081 i P082)	GEN-34A	200
5				Coding sequence analysis with NGS technique	GEN-34C	400
6				Coding sequence analysis - mRNA sequencing **(blood collected on EDTA should be transferred to laboratory in less than 24 hours !!!)**	GEN-34B	550
7	Legius syndrome	*SPRED1*	611431	Coding sequence analysis	GEN-54A	150
8				MLPA test (P295)	GEN-54B	125
9	Noonan syndrome with multiple lentigines (previously LEOPARD syndrome)	*PTPN11*	151100	Analysis of exons: 7, 12, 13	GEN-35A	125
10		*RAF1*	611554	Analysis of exons: 6, 13, 16	GEN-35B	150
11	Aarskog syndrome	*FGD1*	305400	Coding sequence analysis	GEN-52A	250
OTHER CONGENITAL DISORDERS						
1	IRF6 -related disorders (van der Woude syndrome (VWS), Popliteal pterygium syndrome 1 (PPS1))	*IRF6*	119300	Coding sequence analysis	GEN-36A	180
2			119500	Test MLPA (P304)	GEN-36B	125
3	Andersen-Tawil syndrome	*KCNJ2*	170390	Coding sequence analysis	GEN-47A	80
4	Simpson, Golabi Behmel syndrome	*GPC3*	312870	Coding sequence analysis	GEN-51A	190
5	Metaphyseal chondrodysplasia, McKusick type, cartilage-hair hypoplasia, CHH	*RMRP*	250250	Analysis of the entire RNA coding region	GEN-57	80
6	Cowden syndrome / Bannayan-Riley-Ruvalcaba	*PTEN*	158350 153480	Coding sequence analysis	GEN-72A	200
7	Rapp-Hodgkin syndrome	*TP63*	603273	Analysis of exons 13 and 14	GEN-46A	120
8				Analiza pozostałych eksonów genu	GEN-46B	250
9	Neurofibromatosis type II	*NF2*	101000	Analysis of remaining exons of the *NF2* gene	GEN-75A	350

10	Neurofibromatosis type II	*NF2*	101000	MLPA test (P044)	GEN-75B	125
11	Beckwith-Wiedemann syndrome	*11p15*	130650	MS-MLPA test (ME030)	GEN-81A	150
12		*CDKN1C*		Coding sequence analysis	GEN-81E	100
13	Silver-Russell syndrome	*11p15*	180680	MS-MLPA test (ME030)	GEN-81B	150
14		*7p12.1, 7q32.2*		MS-MLPA test (ME032)	GEN-81C	150
15	Multilocus imprinting defects	*multiple loci*		MS-MLPA test (ME034)	GEN-81D	150
16	Transient neonatal diabetes	*6q24*	601410	MS-MLPA test (ME033)	GEN-81F	150
CRANIOSYNOSTOSES AND OTHER FGFR-RELATED DISORDERS						
1	Saethre-Chotzen syndrome	*TWIST1*	101400	Coding sequence analysis	GEN-71A	125
2	Apert syndrome	*FGFR2*	101200	Analysis of exon 7 with identification of p.Ser252Trp and p.Pro253Arg mutations	GEN-71B	80
3	Pfeiffer/Crouzon syndrome	*FGFR2*	101600 123500	Analysis of exons 7 and 8 (8 and 10; identification of most common mutations)	GEN-71C	125
4	Pfeiffer syndrome, type I	*FGFR1*	101600	Analysis of exon 7 with the identification of p.Pro252Arg mutation	GEN-71D	80
5	Muenke syndrome	*FGFR3*	602849	Analysis of exon 7 with the identification of p.Pro250Arg mutation	GEN-71E	80
6	Crozuon syndrome with acanthosis nigricans	*FGFR3*	612247	Analysis of exon 9 with the identification of p.Ala391Glu mutation	GEN-71F	80
7	Achondroplasia	*FGFR3*	100800	Analysis of exon 9 with the identification of p.Gly380Arg mutation	GEN-71G	80
8		*FGFR3*	100800	Analysis of exons: 9,10,11,13,14 i 15	GEN-71J	175
9	Hypochodroplasia	*FGFR3*	146000	Analysis of exon 12 with the identification of p.Asn540Lys mutation	GEN-71H	80
10		*FGFR3*	146000	Analysis of exons: 8,9,13,14 i 15	GEN-71K	150
11	Thanatophoric dysplasia	*FGFR3*		Analysis of exon 7 and 10, including identification of the p.Arg248Cys and p.Tyr373Cys mutations	GEN-71L	125
12	Craniosynostosis - deletion analysis	*nd*	nd	MLPA test (P080)	GEN-71I	125
METABOLIC DISEASES						
1	Phenylketonuria (PKU)	*PAH*	261600	Analysis of exons: 5, 11, 12 [includes detection of: p.Arg408Trp (R408W), c.1066-11G>A (IVS10-11G>A), c.1315+1G>A (IVS12+1G>A), p.Arg158Gln (R158Q)]	GEN-11B	150
2				Analysis of exons:1-4, 6-10, 13	GEN-11C	220
3				MLPA test (P055)	GEN-11D	125
4	Hemochromatosis (HFE)	*HFE*	235200	Detection of mutations p.Cys282Tyr and p.His63Asp	GEN-14	100
5	Lipoid proteinosis (Urbach–Wiethe disease)	*ECM1*	247100	Coding sequence analysis	GEN-58	300
6	Hyperinsulinism/hyperammonemia (HI/HA) syndrome	*GLUD1*	606762	Analysis of exons: 6-12	GEN-59	175
7	Galactosemia (GALT)	*GALT*	230400	Analysis of exons: 6-9 (includes detection of p.Gln188Arg and p.Lys285Asn mutations)	GEN-24A	90
8				Analysis of remaining exons	GEN-24B	120

9				Identification of two mutations - carrier testing	GEN-24C	125
GENODERMATOSIS						
1	Dystrophic epidermolysis bullosa, autosomal dominant (DDEB)	*COL7A1*	131750	Analysis of exons 73-75, includes detection of p.Gly2043Arg	GEN-30E	125
2				Coding sequence analysis	GEN-31B	400
3	Epidermolysis bullosa simplex (SEB) and APSS (acral peeling skin syndrome)	*KRT14, KRT5*	131900 131800 603805	Analysis of KRT5 (exons: 1, 2, 5, 7), KRT14 (exons: 1, 4-7)	GEN-32C	220
4		*KRT14, KRT5, TGM5*		Analysis of remaining exons of KRT5 (exons 3, 4, 6, 8, 9) and KRT14 (exons 2 ,3, 8) with selected exons of TGM5 (exons 2, 3)	GEN-32D	220
5	Epidermolysis bullosa simplex (SEB)	*KRT5*	131900	Coding sequence analysis	GEN-32A	170
6		*KRT14*	131800	Coding sequence analysis	GEN-32B	130
7	APSS (acral peeling skin syndrome)	*TGM5*	603805	Analysis of exons 2, 3	GEN-38A	75
8				Analysis of exons 5, 6, 8, 9	GEN-38B	170
9				Analysis of the remaining exons of the TGM5 gene (1, 4, 7, 10, 11, 12, 13)	GEN-38C	170
10		*CSTA*	184600	Coding sequence analysis	GEN-38D	90
11	Peeling skin syndrome(PSS)	*CDSN*	602593	Coding sequence analysis	GEN-39	125
12	Ichthyosis vulgaris	*FLG*	135940	Identification of the most common mutations: p.Arg501Ter and c.2282_2285del4	GEN-40	100
13	Netherton syndrome	*SPINK5*	605010	Analysis of exons 5, 8, 12-15, 18, 19, 22-26	GEN-42A	320
23				Coding sequence analysis	GEN-42B	450
14	Cloustona syndrome (ectodermal dysplasia)	*GJB6*	604418	Coding sequence analysis, including identification of the p.Gly11Arg and p.Ala88Val mutations	GEN-43	75
15	Hailey-Hailey disease	*ATP2C1*	604384	Analysis of exons: 7, 12, 13, 17, 18, 24, 25	GEN-45A	175
16	Hypohidrotic ectodermal dysplasia (X-linked)	*EDA1*	305100	Analysis of exons 1, 2, 4, 6, 7	GEN-60A	175
17				Analysis of remaining exons (3, 5, 8)	GEN-60B	125
18	X-linked ichthyosis with steryl-sulfatase deficiency	*STS*	308100	MLPA (P160) test - detection of dulications/deletions	GEN-76A	125
19	Pityriasis rubra pilaris	*CARD14*	173200	Analysis of exons: 3, 4, 5	GEN-84A	190
OTHERS						
1	Neural tube defects	*MTHFR*	601634	1298A>C and 677C>T polymorphism analysis	GEN-12	100
2	Thrombosis (inherited thrombophilia, hypercoagulability)	*F2*	188050, 614390	Identification of c.*97G>A (20210G>A) mutation	GEN-56A	75
3		*F5*		Identification of p.Arg534Gln (V Leiden, R506Q) mutation	GEN-56B	75
4		*F2. F5*		Identification of c.*97G>A (20210G>A) mutation in F2 gene and p.Arg534Gln (V Leiden, R506Q) mutation in F5 gene	GEN-56C	100
5		*F2, F5, MTHFR*		Identification of c.*97G>A (20210G>A) mutation in F2 gene, p.Arg534Gln (V Leiden, R506Q) mutation in F5 gene and 677C>T and 1298A>C polymorphisms in MTHFR genes	GEN-56D	150

6	Familial adenomatous polyposis	*APC*	175100	Analysis of 4 most common mutations: c.3927_3931delAAAGA, c3183_3187delACAAA, c.3202_3205delTCAA, p.Tyr500Ter	GEN-70A	140
7	Sex identification	*AMELY, AMELX*	-	Sex determination (amylogenin homologues testing)	GEN-61A	75
9	X chromosome inactivation	*AR*	-	Analysis of X chromosome inactivation	GEN-53A	100
ADDITIONAL INFORMATION						
1	Identification of single mutation (only genes from the offer)	-	-	Sanger sequencing of single gene fragment	GEN-23A	75
2	Identification of any (single) mutation in a gene that is not included in the diagnostic offer (NGS variant verification, carrier test)	-	-	Sanger sequencing of single gene fragment	GEN-23B	120
3	"CITO" analysis	-	-	Offer cost of the analysis + 30%	-	-
4	Prenatal testing	-	-	It is necessary to determine in advance what disease the test will be for and its date		double price
5	Other procedures not included in our offer	-	-	According to the procedure	GEN-16	price to be agreed