



NEXT GENERATION SANGER SEQUENCING PLATFORMS (NGS)

Category	Disease Category	Disease	Number of Genes	Genes	Comment	Test Specification	Price in €
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	COMPREHENSIVE HEREDITARY CANCER	116	AIP, ALK, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CEBPA, CEP57, CHEK2, CYLD, DDB2, DICER1, DIS3L2, DKC1, EGFR, ELANE, EPCAM, ERCC2, ERCC3, ERCC4, ERCC5, EXO1, EXT1, EXT2, EZH2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANGC, FANCI, FANCL, FANCM, FH, FLCN, GATA2, GPC3, GREM1, HNF1A, HOXB13, HRAS, KIT, KRAS, MAX, MEN1, MET, MITF, MLH1, MLH3, MRE11A, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NSD1, PALB2, PHOX2B, PMS1, PMS2, POLD1, POLE, PPM1D, PRF1, PRKAR1A, PTCH1, PTEN, PTPN11, RAD50, RAD51C, RAD51D, RB1, RECQL4, RET, RHBDF2, RUNX1, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLX4, SMAD4, SMARCB1, STK11, SUFU, TERC, TERT, TINF2, TMEM127, TP53, TSC1, TSC2, VHL, WRN, WTI, XPA, XPC, XRCC2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	BREAST, OVARIAN, COLORECTAL CANCER	50	BRCA1, BRCA2, APC, ATM, BAP1, BARD1, BMPR1A, BRIP1, CDH1, CDKN2A, CDK4, CHEK2, EPCAM, GREM1, MLH1, MSH2, MSH6, MITF, MUTYH, NBN, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, SMAD4, STK11, TP53		Sequencing and Deletion-Duplication Testing of all Genes	359
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	BREAST AND OVARIAN CANCER	12	BRCA1, BRCA2, CHEK2, BRIP1, BARD1, CDH1, TP53, PTEN, RAD51C, MRE11A, NBN, ATM		Sequencing	680
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	COLON CANCER	6	APC, MUTYH, MLH1, MSH2, MSH6, PMS1, PMS2, EPCAM, BMPR1A, SMAD4, STK11, PTEN		Sequencing and Deletion-Duplication Testing of all Genes	680
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	GASTROINTESTINAL CANCER	38	APC, ATM, AXIN2, BLM, BMPR1A, BRCA1, BRCA2, BUB1B, CDH1, CDKN2A, EPCAM, FANCC, GREM1, HNF1A, KIT, MEN1, MLH1, MSH2, MSH6, MUTYH, NF1, PALB2, PMS2, POLD1, POLE, PTEN, RHBDF2, SDHB, SDHC, SDHD, SMAD4, SMARCB1, STK11, TMEM127, TP53, TSC1, TSC2, VHL		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	LEUKEMIA	25	ATM, BLM, BRCA2, CDKN2A, CEBPA, DKC1, ELANE, FANCA, GATA2, HRAS, KRAS, MLH1, MSH2, MSH6, NBN, NF1, NRAS, PMS2, PTPN11, RUNX1, SBDS, TERC, TERT, TINF2, TP53		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	LUNG CANCER	4	BRCA2, CDKN2A, EGFR, TP53		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	MELANOMA AND SKIN CANCER	19	BAP1, BRCA1, BRCA2, CDK4, CDKN2A, DDB2, ERCC2, ERCC3, ERCC4, ERCC5, MITF, PTCH1, PTEN, RB1, SILE1, TP53, WRN, XPA, XPC		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	PANCREATIC CANCER	22	APC, ATM, BMPR1A, BRCA1, BRCA2, BUB1B, CDKN2A, EPCAM, FANCC, MEN1, MLH1, MSH2, MSH6, NF1, PALB2, PMS2, SMAD4, STK11, TP53, TSC1, TSC2, VHL		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	PARAGANGLIOMA-PHEOCHROMOCYTOMA	11	FH, MAX, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	PEDIATRIC CANCER	52	ALK, APC, AXIN2, BAP1, BLM, BMPR1A, CDC73, CDKN1C, CEBPA, DICER1, DIS3L2, EPCAM, EZH2, FH, GATA2, GPC3, HRAS, MAX, MEN1, MLH1, MSH2, MSH6, NBN, NF1, NF2, NSD1, PHOX2B, PMS2, PRF1, PRKAR1A, PTCH1, PTEN, RB1, RECQL4, RET, RUNX1, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCB1, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL, WRN, XPA, XPC		Sequencing and Deletion-Duplication Testing of all Genes	
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	RENAL CANCER (PANEL 1)	25	BAP1, CDC73, CDKN1C, DICER1, DIS3L2, EPCAM, FH, FLCN, GPC3, HNF1A, MET, MLH1, MSH2, MSH6, PMS2, PTEN, SDHB, SDHC, SDHD, SMARCB1, TP53, TSC1, TSC2, VHL, WTI		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	RENAL CANCER (PANEL 2)	19	BAP1, FH, FLCN, MET, MITF, MLH1, MSH2, MSH6, PMS2, PTEN, SDHA, SDHB, SDHC, SDHD, TP53, TSC1, TSC2, VHL, EPCAM (Deletion/Duplication Testing only)		Sequencing of 18 Genes and Deletion-Duplication Testing of 19 Genes	1850
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	THYROID CANCER	11	APC, CDC73, DICER1, MEN1, PRKAR1A, PTEN, RET, SDHB, SDHD, TP53, WRN		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	NEUROFIBROMATOSIS	7	KIT, NF1, NF2, PTPN11, RAF1, SMARCB1, SPRED1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	TUBEROUS SCLEROSIS	2	TSC1, TSC2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	XERODERMA PIGMENTOSUM (PANEL 1)	7	DDB2, ERCC2, ERCC3, ERCC4, ERCC5, XPA, XPC		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	XERODERMA PIGMENTOSUM (PANEL 2)	9	DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC		Sequencing	1750
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	COMPREHENSIVE CARDIOLOGY	165	A2ML1, AARS2, ABCB4, ABCG9, ACAD9, ACADVL, ACTA1, ACTA2, ACTC1, ACTN2, AGK, AGL, AKAP9, ALPK3, ANK2, ANKRD1, ANOS, APOA1, APOB, BAG3, BRAF, CACNA1C, CACNA2D4, CACNB2, CALM1, CALM2, CALM3, CAPN3, CASQ2, CAV3, CBL, CHKB, COX15, CPT1A, CPT2, CRYAB, CSRP3, CTNNA3, DAG1, DBH, DES, DMD, DNAIC19, DSC2, DSG2, DSP, DYSF, EMD, ENPP1, ETFA, ETFB, ETFDH, EYA4, FBXO32, FHL1, FKRP, FKTN, FLNC, FOXRED1, FXN, GAA, GATAS, GATAD1, GBE1, GFM1, GJA5, GLA, GLB1, GMPBB, GNE, GUSB, HCN4, HFE, HRAS, ISPD, JPH2, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNQ1, KRAS, LAMA2, LAMP2, LARGE, LDB3, LMNA, LZTR1, MAP2K1, MAP2K2, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYOT, MYPN, NEXN, NF1, NKX2-5, NOS1AP, NRAS, NSUN2, PKP2, PLEC, PLEKHM2, PLN, PNPLA2, POMGNT1, POMT1, POMT2, PRDM16, PRKAG2, PTPN11, RAF1, RASA2, RBM20, RIT1, RRAS, RYR2, SCN10A, SCN1B, SCN3B, SCN5A, SCN1B, SCNN1G, SCO2, SDHA, SELENON, SGCA, SGCB, SGCD, SGCG, SHOC2, SLC22A5, SLC25A20, SLC25A4, SMCHD1, SOS1, SPRED1, TAZ, TBX5, TCAP, TGFB3, TMEM43, TMEM70, TNNC1, TNNT3, TNNT2, TPML1, TRDN, TRIM32, TRPM4, TSFM, TTN, TCFE7L2, TCFE7L3, TCFE7L4, TCFE7L5, TCFE7L6, TCFE7L7, TCFE7L8, TCFE7L9, TCFE7L10, TCFE7L11, TCFE7L12, TCFE7L13, TCFE7L14, TCFE7L15, TCFE7L16, TCFE7L17, TCFE7L18, TCFE7L19, TCFE7L20, TCFE7L21, TCFE7L22, TCFE7L23, TCFE7L24, TCFE7L25, TCFE7L26, TCFE7L27, TCFE7L28, TCFE7L29, TCFE7L30, TCFE7L31, TCFE7L32, TCFE7L33, TCFE7L34, TCFE7L35, TCFE7L36, TCFE7L37, TCFE7L38, TCFE7L39, TCFE7L40, TCFE7L41, TCFE7L42, TCFE7L43, TCFE7L44, TCFE7L45, TCFE7L46, TCFE7L47, TCFE7L48, TCFE7L49, TCFE7L50, TCFE7L51, TCFE7L52, TCFE7L53, TCFE7L54, TCFE7L55, TCFE7L56, TCFE7L57, TCFE7L58, TCFE7L59, TCFE7L60, TCFE7L61, TCFE7L62, TCFE7L63, TCFE7L64, TCFE7L65, TCFE7L66, TCFE7L67, TCFE7L68, TCFE7L69, TCFE7L70, TCFE7L71, TCFE7L72, TCFE7L73, TCFE7L74, TCFE7L75, TCFE7L76, TCFE7L77, TCFE7L78, TCFE7L79, TCFE7L80, TCFE7L81, TCFE7L82, TCFE7L83, TCFE7L84, TCFE7L85, TCFE7L86, TCFE7L87, TCFE7L88, TCFE7L89, TCFE7L90, TCFE7L91, TCFE7L92, TCFE7L93, TCFE7L94, TCFE7L95, TCFE7L96, TCFE7L97, TCFE7L98, TCFE7L99, TCFE7L100, TCFE7L101, TCFE7L102, TCFE7L103, TCFE7L104, TCFE7L105, TCFE7L106, TCFE7L107, TCFE7L108, TCFE7L109, TCFE7L110, TCFE7L111, TCFE7L112, TCFE7L113, TCFE7L114, TCFE7L115, TCFE7L116, TCFE7L117, TCFE7L118, TCFE7L119, TCFE7L120, TCFE7L121, TCFE7L122, TCFE7L123, TCFE7L124, TCFE7L125, TCFE7L126, TCFE7L127, TCFE7L128, TCFE7L129, TCFE7L130, TCFE7L131, TCFE7L132, TCFE7L133, TCFE7L134, TCFE7L135, TCFE7L136, TCFE7L137, TCFE7L138, TCFE7L139, TCFE7L140, TCFE7L141, TCFE7L142, TCFE7L143, TCFE7L144, TCFE7L145, TCFE7L146, TCFE7L147, TCFE7L148, TCFE7L149, TCFE7L150, TCFE7L151, TCFE7L152, TCFE7L153, TCFE7L154, TCFE7L155, TCFE7L156, TCFE7L157, TCFE7L158, TCFE7L159, TCFE7L160, TCFE7L161, TCFE7L162, TCFE7L163, TCFE7L164, TCFE7L165, TCFE7L166, TCFE7L167, TCFE7L168, TCFE7L169, TCFE7L170, TCFE7L171, TCFE7L172, TCFE7L173, TCFE7L174, TCFE7L175, TCFE7L176, TCFE7L177, TCFE7L178, TCFE7L179, TCFE7L180, TCFE7L181, TCFE7L182, TCFE7L183, TCFE7L184, TCFE7L185, TCFE7L186, TCFE7L187, TCFE7L188, TCFE7L189, TCFE7L190, TCFE7L191, TCFE7L192, TCFE7L193, TCFE7L194, TCFE7L195, TCFE7L196, TCFE7L197, TCFE7L198, TCFE7L199, TCFE7L200, TCFE7L201, TCFE7L202, TCFE7L203, TCFE7L204, TCFE7L205, TCFE7L206, TCFE7L207, TCFE7L208, TCFE7L209, TCFE7L210, TCFE7L211, TCFE7L212, TCFE7L213, TCFE7L214, TCFE7L215, TCFE7L216, TCFE7L217, TCFE7L218, TCFE7L219, TCFE7L220, TCFE7L221, TCFE7L222, TCFE7L223, TCFE7L224, TCFE7L225, TCFE7L226, TCFE7L227, TCFE7L228, TCFE7L229, TCFE7L230, TCFE7L231, TCFE7L232, TCFE7L233, TCFE7L234, TCFE7L235, TCFE7L236, TCFE7L237, TCFE7L238, TCFE7L239, TCFE7L240, TCFE7L241, TCFE7L242, TCFE7L243, TCFE7L244, TCFE7L245, TCFE7L246, TCFE7L247, TCFE7L248, TCFE7L249, TCFE7L250, TCFE7L251, TCFE7L252, TCFE7L253, TCFE7L254, TCFE7L255, TCFE7L256, TCFE7L257, TCFE7L258, TCFE7L259, TCFE7L260, TCFE7L261, TCFE7L262, TCFE7L263, TCFE7L264, TCFE7L265, TCFE7L266, TCFE7L267, TCFE7L268, TCFE7L269, TCFE7L270, TCFE7L271, TCFE7L272, TCFE7L273, TCFE7L274, TCFE7L275, TCFE7L276, TCFE7L277, TCFE7L278, TCFE7L279, TCFE7L280, TCFE7L281, TCFE7L282, TCFE7L283, TCFE7L284, TCFE7L285, TCFE7L286, TCFE7L287, TCFE7L288, TCFE7L289, TCFE7L290, TCFE7L291, TCFE7L292, TCFE7L293, TCFE7L294, TCFE7L295, TCFE7L296, TCFE7L297, TCFE7L298, TCFE7L299, TCFE7L300, TCFE7L301, TCFE7L302, TCFE7L303, TCFE7L304, TCFE7L305, TCFE7L306, TCFE7L307, TCFE7L308, TCFE7L309, TCFE7L310, TCFE7L311, TCFE7L312, TCFE7L313, TCFE7L314, TCFE7L315, TCFE7L316, TCFE7L317, TCFE7L318, TCFE7L319, TCFE7L320, TCFE7L321, TCFE7L322, TCFE7L323, TCFE7L324, TCFE7L325, TCFE7L326, TCFE7L327, TCFE7L328, TCFE7L329, TCFE7L330, TCFE7L331, TCFE7L332, TCFE7L333, TCFE7L334, TCFE7L335, TCFE7L336, TCFE7L337, TCFE7L338, TCFE7L339, TCFE7L340, TCFE7L341, TCFE7L342, TCFE7L343, TCFE7L344, TCFE7L345, TCFE7L346, TCFE7L347, TCFE7L348, TCFE7L349, TCFE7L350, TCFE7L351, TCFE7L352, TCFE7L353, TCFE7L354, TCFE7L355, TCFE7L356, TCFE7L357, TCFE7L358, TCFE7L359, TCFE7L360, TCFE7L361, TCFE7L362, TCFE7L363, TCFE7L364, TCFE7L365, TCFE7L366, TCFE7L367, TCFE7L368, TCFE7L369, TCFE7L370, TCFE7L371, TCFE7L372, TCFE7L373, TCFE7L374, TCFE7L375, TCFE7L376, TCFE7L377, TCFE7L378, TCFE7L379, TCFE7L380, TCFE7L381, TCFE7L382, TCFE7L383, TCFE7L384, TCFE7L385, TCFE7L386, TCFE7L387, TCFE7L388, TCFE7L389, TCFE7L390, TCFE7L391, TCFE7L392, TCFE7L393, TCFE7L394, TCFE7L395, TCFE7L396, TCFE7L397, TCFE7L398, TCFE7L399, TCFE7L400, TCFE7L401, TCFE7L402, TCFE7L403, TCFE7L404, TCFE7L405, TCFE7L406, TCFE7L407, TCFE7L408, TCFE7L409, TCFE7L410, TCFE7L411, TCFE7L412, TCFE7L413, TCFE7L414, TCFE7L415, TCFE7L416, TCFE7L417, TCFE7L418, TCFE7L419, TCFE7L420, TCFE7L421, TCFE7L422, TCFE7L423, TCFE7L424, TCFE7L425, TCFE7L426, TCFE7L427, TCFE7L428, TCFE7L429, TCFE7L430, TCFE7L431, TCFE7L432, TCFE7L433, TCFE7L434, TCFE7L435, TCFE7L436, TCFE7L437, TCFE7L438, TCFE7L439, TCFE7L440, TCFE7L441, TCFE7L442, TCFE7L443, TCFE7L444, TCFE7L445, TCFE7L446, TCFE7L447, TCFE7L448, TCFE7L449, TCFE7L450, TCFE7L451, TCFE7L452, TCFE7L453, TCFE7L454, TCFE7L455, TCFE7L456, TCFE7L457, TCFE7L458, TCFE7L459, TCFE7L460, TCFE7L461, TCFE7L462, TCFE7L463, TCFE7L464, TCFE7L465, TCFE7L466, TCFE7L467, TCFE7L468, TCFE7L469, TCFE7L470, TCFE7L471, TCFE7L472, TCFE7L473, TCFE7L474, TCFE7L475, TCFE7L476, TCFE7L477, TCFE7L478, TCFE7L479, TCFE7L480, TCFE7L481, TCFE7L482, TCFE7L483, TCFE7L484, TCFE7L485, TCFE7L486, TCFE7L487, TCFE7L488, TCFE7L489, TCFE7L490, TCFE7L491, TCFE7L492, TCFE7L493, TCFE7L494, TCFE7L495, TCFE7L496, TCFE7L497, TCFE7L498, TCFE7L499, TCFE7L500, TCFE7L501, TCFE7L502, TCFE7L503, TCFE7L504, TCFE7L505, TCFE7L506, TCFE7L507, TCFE7L508, TCFE7L509, TCFE7L510, TCFE7L511, TCFE7L512, TCFE7L513, TCFE7L514, TCFE7L515, TCFE7L516, TCFE7L517, TCFE7L518, TCFE7L519, TCFE7L520, TCFE7L521, TCFE7L522, TCFE7L523, TCFE7L524, TCFE7L525, TCFE7L526, TCFE7L527, TCFE7L528, TCFE7L529, TCFE7L530, TCFE7L531, TCFE7L532, TCFE7L533, TCFE7L534, TCFE7L535, TCFE7L536, TCFE7L537, TCFE7L538, TCFE7L539, TCFE7L540, TCFE7L541, TCFE7L542, TCFE7L543, TCFE7L544, TCFE7L545, TCFE7L546, TCFE7L547, TCFE7L548, TCFE7L549, TCFE7L550, TCFE7L551, TCFE7L552, TCFE7L553, TCFE7L554, TCFE7L555, TCFE7L556, TCFE7L557, TCFE7L558, TCFE7L559, TCFE7L560, TCFE7L561, TCFE7L562, TCFE7L563, TCFE7L564, TCFE7L565, TCFE7L566, TCFE7L567, TCFE7L568, TCFE7L569, TCFE7L570, TCFE7L571, TCFE7L572, TCFE7L573, TCFE7L574, TCFE7L575, TCFE7L576, TCFE7L577, TCFE7L578, TCFE7L579, TCFE7L580, TCFE7L581, TCFE7L582, TCFE7L583, TCFE7L584, TCFE7L585, TCFE7L586, TCFE7L587, TCFE7L588, TCFE7L589, TCFE7L590, TCFE7L591, TCFE7L592, TCFE7L593, TCFE7L594, TCFE7L595, TCFE7L596, TCFE7L597, TCFE7L598, TCFE7L599, TCFE7L600, TCFE7L601, TCFE7L602, TCFE7L603, TCFE7L604, TCFE7L605, TCFE7L606, TCFE7L607, TCFE7L608, TCFE7L609, TCFE7L610, TCFE7L611, TCFE7L612, TCFE7L613, TCFE7L614, TCFE7L615, TCFE7L616, TCFE7L617, TCFE7L618, TCFE7L619, TCFE7L620, TCFE7L621, TCFE7L622, TCFE7L623, TCFE7L624, TCFE7L625, TCFE7L626, TCFE7L627, TCFE7L628, TCFE7L629, TCFE7L630, TCFE7L631, TCFE7L632, TCFE7L633, TCFE7L634, TCFE7L635, TCFE7L636, TCFE7L637, TCFE7L638, TCFE7L639, TCFE7L640, TCFE7L641, TCFE7L642, TCFE7L643, TCFE7L644, TCFE7L645, TCFE7L646, TCFE7L647, TCFE7L648, TCFE7L649, TCFE7L650, TCFE7L651, TCFE7L652, TCFE7L653, TCFE7L654, TCFE7L655, TCFE7L656, TCFE7L657, TCFE7L658, TCFE7L659, TCFE7L660, TCFE7L661, TCFE7L662, TCFE7L663, TCFE7L664, TCFE7L665, TCFE7L666, TCFE7L667, TCFE7L668, TCFE7L669, TCFE7L670, TCFE7L671, TCFE7L672, TCFE7L673, TCFE7L674, TCFE7L675, TCFE7L676, TCFE7L677, TCFE7L678, TCFE7L679, TCFE7L680, TCFE7L681, TCFE7L682, TCFE7L683, TCFE7L684, TCFE7L685, TCFE7L686, TCFE7L687, TCFE7L688, TCFE7L689, TCFE7L690, TCFE7L691, TCFE7L692, TCFE7L693, TCFE7L694, TCFE7L695, TCFE7L696, TCFE7L697, TCFE7L698, TCFE7L699, TCFE7L700, TCFE7L701, TCFE7L702, TCFE7L703, TCFE7L704, TCFE7L705, TCFE7L706, TCFE7L707, TCFE7L708, TCFE7L709, TCFE7L710, TCFE7L711, TCFE7L712, TCFE7L713, TCFE7L714, TCFE7L715, TCFE7L716, TCFE7L717, TCFE7L718, TCFE7L719, TCFE7L720, TCFE7L721, TCFE7L722, TCFE7L723, TCFE7L724, TCFE7L725, TCFE7L726, TCFE7L727, TCFE7L728, TCFE7L729, TCFE7L730, TCFE7L731, TCFE7L732, TCFE7L733, TCFE7L734, TCFE7L735, TCFE7L736, TCFE7L737, TCFE7L738, TCFE7L739, TCFE7L740, TCFE7L741, TCFE7L742, TCFE7L743, TCFE7L744, TCFE7L745, TCFE7L746, TCFE7L747, TCFE7L748, TCFE7L749, TCFE7L750, TCFE7L751, TCFE7L752, TCFE7L753, TCFE7L754, TCFE7L755, TCFE7L756, TCFE7L757, TCFE7L758, TCFE7L759, TCFE7L760, TCFE7L761, TCFE7L762, TCFE7L763, TCFE7L764, TCFE7L765, TCFE7L766, TCFE7L767, TCFE7L768, TCFE7L769, TCFE7L770, TCFE7L771, TCFE7L772, TCFE7L773, TCFE7L774, TCFE7L775, TCFE7L776, TCFE7L777, TCFE7L778, TCFE7L779, TCFE7L780, TCFE7L781, TCFE7L782, TCFE7L783, TCFE7L784, TCFE7L785, TCFE7L786, TCFE7L787, TCFE7L788, TCFE7L789, TCFE7L790, TCFE7L791, TCFE7L792, TCFE7L793, TCFE7L794, TCFE7L795, TCFE7L796, TCFE7L797, TCFE7L798, TCFE7L799, TCFE7L800, TCFE7L801, TCFE7L802, TCFE7L803, TCFE7L804, TCFE7L805, TCFE7L806, TCFE7L807, TCFE7L808, TCFE7L809, TCFE7L810, TCFE7L811, TCFE7L812, TCFE7L813, TCFE7L814, TCFE7L815, TCFE7L816, TCFE7L817, TCFE7L818, TCFE7L819, TCFE7L820, TCFE7L821, TCFE7L822, TCFE7L823, TCFE7L824, TCFE7L825, TCFE7L826, TCFE7L827, TCFE7L828, TCFE7L829, TCFE7L830, TCFE7L831, TCFE7L832, TCFE7L833, TCFE7L834, TCFE7L835, TCFE7L836, TCFE7L837, TCFE7L838, TCFE7L839, TCFE7L840, TCFE7L841, TCFE7L842, TCFE7L843, TCFE7L844, TCFE7L845, TCFE7L846, TCFE7L847, TCFE7L848, TCFE7L849, TCFE7L850, TCFE7L851, TCFE7L852, TCFE7L853, TCFE7L854, TCFE7L855, TCFE7L856, TCFE7L857, TCFE7L858, TCFE7L859, TCFE7L860, TCFE7L861, TCFE7L862, TCFE7L863, TCFE7L864, TCFE7L865, TCFE7L866, TCFE7L867, TCFE7L868, TCFE7L869, TCFE7L870, TCFE7L871, TCFE7L872, TCFE7L873, TCFE7L874, TCFE7L875, TCFE7L876, TCFE7L877, TCFE7L878, TCFE7L879, TCFE7L880, TCFE7L881, TCFE7L882, TCFE7L883, TCFE7L884, TCFE7L885, TCFE7L886, TCFE7L887, TCFE7L888, TCFE7L889, TCFE7L890, TCFE7L891, TCFE7L892, TCFE7L893, TCFE7L894, TCFE7L895, TCFE7L896, TCFE7L897, TCFE7L898, TCFE7L899, TCFE7L900, TCFE7L901, TCFE7L902, TCFE7L903, TCFE7L904, TCFE7L905, TCFE7L906, TCFE7L907, TCFE7L908, TCFE7L909, TCFE7L910, TCFE7L911, TCFE7L912, TCFE7L913, TCFE7L914, TCFE7L915, TCFE7L916, TCFE7L917, TCFE7L918, TCFE7L919, TCFE7L920, TCFE7L921, TCFE7L922, TCFE7L923, TCFE7L924, TCFE7L925			

Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	ALBINISM	19	AP3B1, BLOC1S3, BLOC1S6, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, MC1R, MITF, MYO5A, OCA2, RAB27A, SLC45A2, TYR, TYRP1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	CUTIS LAXA	8	ALDH18A1, ATP4A, ATP6V0A2, EFEMP2, ELN, FBLN5, PYCR1, SLC2A10	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	CUTIS LAXA (PANEL 2)	13	ELN, FBLN4 (EFEMP2), FBLN5, LTBPA, ATP6V0A2, PYCR1, ALDH18A1, ATP7A, COG7, TAI1D01, SCY11R1P1 (GORAB), NAA10, RIN2	Sequencing	1600
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	DYSKERATOSIS CONGENITA	11	AK2, CTC1, DKC1, NHP2, NOP10, RTE1L, TERC, TERT, TINF2, USB1, WRAP53	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	ECTODERMAL DYSPLASIA	19	ABCC9, BCS1L, DSP, EDA, EDAR, ERCC2, EVC, EVC2, GJB2, GJB6, IFT122, IKBKG, JUP, PORCN, RMRP, SHOC2, TP63, WDR35, WNT10A	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	EHLERS-DANLOS SYNDROME	33	ABCC6, ADAMTS2, ALDH18A1, ATP6V0A2, ATP7A, CBS, CHST14, COL11A1, COL18A1, COL11A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, PLOD1, PYCR1, SLC39A13, SMAD3, TGFB2, TGFB3, TGFB3L1, TRAP1, TRAP2, TRAP3, TRAP4	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	EPIDERMOLYSIS BULLOSA	25	ATP2C1, CDSN, COL17A1, COL7A1, DSG1, DSG2, DSG4, DSP, DST, EXPH5, FERMT1, GRIP1, ITGA3, ITGA6, ITGB4, KRT1, KRT14, KRT5, LAMA3, LAMB3, LAMC2, MMP1, PKP1, PLEC, TGM5	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	EPIDERMOLYSIS BULLOSA (PANEL 2)	21	CD151, CDSN, CHST8, COL7A1, COL17A1, DSP, DST, EXPH5, FERMT1, ITGA3, ITGA6, ITGB4, JUP, KRT5, KRT14, LAMA3, LAMB3, LAMC2, PKP1, PLEC1, TGM5	Sequencing	1300
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	HERMANSKY-PUDLAK SYNDROME	22	ABCA3, AP3B1, BLOC1S3, BLOC1S6, DKC1, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, OCA2, SFTPB, SFTPC, SLC45A2, TERC, TERT, TINF2, TYR, TYRP1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	ICHTHYOSIS	19	ABCA12, ALG8, ALOX12B, ALOXE3, CYP4F22, EBP, ERCC2, FLG, GJA1, GJB2, GJB3, KRT1, KRT2, KRT9, PEX7, PHVH, STS, SUIMF1, TGM1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	NEUROFIBROMATOSIS	7	KIT, NF1, NF2, PTPN11, RAF1, SMARCB1, SPRED1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	PACHYONYCHIA CONGENITA	8	AAGAB, GJB6, KRT16, KRT17, KRT6A, KRT6B, KRT6C, TRPV3	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	PROGERIA AND PROGEROID SYNDROMES	16	AGPAT2, ALDH18A1, B4GALT7, BLM, BSLC2, COL3A1, ERCC2, ERCC4, ERCC5, ERCC6, ERCC8, LMNA, PYCR1, RECQL4, WRN, ZMPSTE24	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	WAARDENBURG SYNDROME	6	EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-Nose-Throat	COMPREHENSIVE HEARING LOSS	158	ABHD12, ACTG1, ADCY1, ADGRV1, ALMS1, ANKH, ATP6V1B1, BCS1L, BDP1, BSND, BTD, CABP2, CACNA1D, CCDC50, CD151, CDH23, CDKN1C, CEACAM16, CHD7, CHSY1, CIB2, CLDN14, CLIC5, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DCDC2, DFNAS, DFNB31, DFNBS9, DIABLO, DIAPH1, DIAPH3, DLX5, DSPP, EDN3, EDNRB, ELMOD3, EPS8, ESPN, ESRRB, EYA1, EYA4, FAM65B, FGF3, FGF3L, FOXI1, GATA3, GIPC3, GJB2, GJB3, GJB6, GPM2, GRHL2, GRXCR1, GRXCR2, HARS, HGF, HOMER2, HOXB1, HSD17B4, ILDR1, KARS, KCNE1, KCNQ1, KCNQ4, LHFPL5, LOXHD1, LRP2, LRTOMT, MANBA, MARVELD2, MET, MIR96, MITF, MSRB3, MYH14, MYH9, MYO15A, MYO1A, MYO3A, MYO6, MYO7A, NARS2, NDP, NLRP3, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDZD7, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, PTPRO, RDX, RMND1, RPS6KA3, SALL4, SEMA3E, SERPINB6, SIX1, SIX5, SLC17A8, SLC19A2, SLC26A4, SLC26A5, SLC29A3, SLC33A1, SLITRK6, SMAD4, SMPX, SNAI2, SOX10, STRC, SUCLA2, SUCLG1, SYNE4, TBC1D24, TCOF1, TECTA, TFAP2A, TIMM8A, TJP2, TMC1, TMC2, TMEM132E, TMIE, TMPRSS3, TNC, TRPN, TROBP, TRMU, TSPEAR, TYR, USH1C, USH1G, USH2A, VCAN, WFS1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-Nose-Throat	ALPURT SYNDROME	6	CD151, COL4A3, COL4A4, COL4A6, MYH5	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-Nose-Throat	BRANCHIO-OTO-RENAL (BOR) SYNDROME	4	EYA1, SIX1, SIX5, TFAP2A	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-Nose-Throat	HEREDITARY HEMORRHAGIC TELANGIECTASIA (HHT)	4	ACVRL1, ENG, RASA1, SMAD4	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-Nose-Throat	NON-SYNDROMIC HEARING LOSS	93	ACTG1, ADCY1, BDP1, BSND, CABP2, CCDC50, CDH23, CEACAM16, CIB2, CLDN14, CLIC5, COCH, COL11A2, COL4A6, CRYM, DCDC2, DFNAS, DFNB31, DFNBS9, DIABLO, DIAPH1, DIAPH3, DSPP, ELMOD3, EPS8, ESPN, ESRRB, EYA4, FAM65B, GIPC3, GJB2, GJB3, GJB6, GPM2, GRHL2, GRXCR1, GRXCR2, HGF, HOMER2, ILDR1, KARS, KCNQ4, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MET, MIR96, MSRB3, MYH14, MYH9, MYO15A, MYO1A, MYO3A, MYO6, MYO7A, NARS2, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PCDH15, PNPT1, POU3F4, POU4F3, PRPS1, PTPRO, RDX, SERPINB6, SIX1, SLC17A8, SLC26A4, SLC26A5, SLITRK6, SMPX, STRC, SYNE4, TBC1D24, TECTA, TJP2, TMC1, TMC2, TMEM132E, TMIE, TMPRSS3, TNC, TRPN, TROBP, TSPEAR, USH1C, WFS1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-Nose-Throat	PENDRED SYNDROME	3	FOX11, KCNQ1, SLC26A4	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-Nose-Throat	SYNDROMIC HEARING LOSS	70	ABHD12, ACTG1, ADGRV1, ALMS1, ANKH, ATP6V1B1, BCS1L, BSND, BTD, CACNA1D, CD151, CDH23, CDKN1C, CHD7, CHSY1, CIB2, CLRN1, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, DFNB31, DLX5, EDN3, EDNRB, EYA1, FGF3, FOXI1, GATA3, HARS, HOXB1, KCNE1, KCNQ1, KCNQ4, LRP2, MANBA, MITF, MYH9, MYO7A, NDP, NLRP3, PAX3, PCDH15, PDZD7, POLR1C, POLR1D, SEMA3E, SIX1, SIX5, SLC19A2, SLC26A4, SLITRK6, SMAD4, SNAI2, SOX10, TCOF1, TFAP2A, TIMM8A, TYR, USH1C, USH1G, USH2A, VCAN, WFS1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-Nose-Throat	USHER SYNDROME	13	ABHD12, ADGRV1, CDH23, CIB2, CLRN1, DFNB31, HARS, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-Nose-Throat	WAARDENBURG SYNDROME	6	EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	ABNORMAL GENITALIA/ DISORDERS OF SEX DEVELOPMENT	39	AMH, AMHR2, ANOS1, AR, ARX, ATRX, BCOR, CDKN1C, CEP41, CHD7, CREBBP, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DYNC2H1, FIG4, FRAS1, GATA4, GNRHR, HSD17B3, HSD3B2, IL17RD, IRF6, LHCGR, MKS1, NR0B1, NR5A1, POR, PROKR2, RSP01, SOX9, SPO11, SRY, STAR, TAC3, WFI, ZFN1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	CONGENITAL ADRENAL HYPERPLASIA	7	CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, PRKAR1A, STAR	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	GLUCOCORTICOID DEFICIENCY	6	MC2R, MRAP, NNT, NR3C1, POMC, STAR	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	HYPERPARATHYROIDISM	6	AIRE, CASR, CDC73, CDKN1B, MEN1, RET	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	HYPOGLYCEMIA, HYPERINSULINISM AND KETONE METABOLISM	18	ABCC8, ACAT1, FBP1, GCK, GLUD1, HADH, HMGCCL, HMGCS2, HNF1A, HNF4A, INSR, KCN11, OXCT1, PCK1, PCK2, PDX1, SLC16A1, UCP2	Sequencing and Deletion-Duplication Testing of all Genes	1290

Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	HYPOMAGNESEMIA	17	CLDN16, CLDN19, CNNM1, CNNM2, CNNM4, EGF, FXYP2, HNF1B, KCNA1, MAGT1, MMTG1, NIPA2, SLC12A3, SLC41A2, SLC41A3, TRPM6, TRPM7	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	HYPOTHYROIDISM AND RESISTANCE TO THYROID HORMONE	17	DUOX2, GNAS, HESX1, NKX2-1, NKX2-5, PAX8, POU1F1, PROPI, SLC16A2, SLC26A4, SLC5A5, TG, THRA, THRB, TPO, TSHR, TSHR	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	KALLMANN SYNDROME	8	ANOS1, CHD7, FGF8, FGF81, GNRHR, PROK2, PROKR2, TACR3	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	MODY	12	ABCC8, BLK, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	MONOGENIC DIABETES MELLITUS	25	ABCC8, BLK, EIF2AK3, FOXP3, G6PC2, GCK, GLIS3, GLUD1, HADH, HNF1A, HNF1B, HNF4A, INS, INSR, KCNJ11, KLF11, NEUROD1, NEUROG3, PAX4, PDX1, PPARG, SLC16A1, SLC2A2, UCP2, WFS1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	MONOGENIC OBESITY	36	ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, CRT1, CUL4B, DYRK1B, GNAS, LEP, LEPR, MAGEL2, MC3R, MC4R, MKKS, MKS1, NROB2, NTRK2, PCSK1, PHF6, POMC, PPARG, PYY, SDCCAG8, SIM1, TRIM32, TTC8, UCP3, VPS13B, WDPCP	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	PREMATURE OVARIAN FAILURE	13	CYP17A1, CYP19A1, FOXL2, FSHR, GALT, GNAS, LHCGR, LMNA, NR5A1, POLG, POR, STAR, WT1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	PREMATURE OVARIAN FAILURE (PANEL 2)	17	NOBOX, LHR, STAR, BMP15, PSMC3IP, WT1, DIAPH2, FSHB, GDF9, FOXL2, WNT4, FSHR, FIGL1, CYP17A1, CYP19A1, POF1B	Sequencing of 16 genes and repeat FMR1	2800
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	CHOLESTASIS	31	ABCB11, ABCB4, ABCC2, ATP8B1, CFTR, CYP7B1, DGUOK, EPCAM, FAH, JAG1, LCT, MKS1, MYO5B, NEUROG3, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, SERPINA1, SLC25A13, SLC26A3, SMPD1, SPINT2, TJP2, TMEM216, TRMU, TTC37, UGT1A1, VPS33B	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	CHOLESTASIS (PANEL 2)	15	ABCB11, ABCB4, AKR1D1, ATP8B1, BAAT, CLDN1, HSD3B7, JAG1, NOTCH2, NR1H4, SERPINA1, SLC25A13, TJP2, VIPAS39, VPS33B	Sequencing	1700
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	CONGENITAL DIARRHEA	9	DGAT1, EPCAM, GUCY2C, LCT, MYO5B, NEUROG3, SLC26A3, SPINT2, TTC37	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	CONGENITAL HEPATIC FIBROSIS	29	AH1, ARL13B, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CC2D2A, CEP290, INVS, IQCB1, MKKS, MKS1, NPHP1, NPHP3, NPHP4, OFD1, PKD2, PKHD1, RRGPR1, TMEM67, TRIM33, TTC31B, TTC8, WDOR3	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	GASTROINTESTINAL ATRESIA	7	CHD7, FANCB, FANCC, GLI3, MID1, SOX2, TTC7A	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	HIRSCHSPRUNG DISEASE	9	EDN3, EDNRB, KIF18P, MIF, NRG1, PAX3, RET, SOX10, ZEB2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	PANCREATITIS	5	CFTR, CPA1, CTSC, PRSS1, SPINK1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	POLYCYSTIC LIVER DISEASE	5	LRPS, PKD1, PKD2, PRKCSH, SEC63	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	COMPREHENSIVE HEMATOLOGY	175	ABCA3, ABCB7, ABCG5, ABCG8, ACTB, ACTN1, ADAMTS13, AK2, ALAS2, AMN, ANK1, ANKRD26, AP3B1, ATM, ATR, ATRX, BLM, BLOC1S3, BLOC1S6, BRCA2, BRIP1, C15ORF41, CDAN1, CDKN2A, CEBPA, CSF2RA, CTC1, CTSC, CUBN, CXCR4, CYCS, DKC1, DTNBP1, ELANE, EPB42, ERCC4, F10, F11, F12, F13A1, F2, F5, F7, F8, F9, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FGA, FGB, FGG, FLNA, G6PC3, G6PD, GATA1, GATA2, GGCX, GP1BA, GP1BB, GP9, GPI, GPR143, GSS, HAX1, HBA1, HBA2, HBB, HFE, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, IFNGR2, ITGA2, ITGA2B, ITGB3, ITK, JAGN1, KLF1, KRAS, LMNA, LPIN2, LYST, MAGT1, MASTL, MLH1, MPL, MSH2, MSH6, MTR, MYH9, MYO5A, NBEAL2, NBN, NF1, NHP2, NOP10, NRAS, OCA2, P2RY12, PALB2, PC, PDHA1, PDHX, PKLR, PMS2, PRF1, PROC, PROS1, PTPN11, PUS1, RAB27A, RADS1C, RBM8A, RECQL4, RPL11, RPL15, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS29, RPS7, RTEL1, RUNX1, SBDS, SEC23B, SERPINC1, SFTPB, SFTPC, SH2D1A, SLC19A2, SLC45A2, SLC4A1, SLFN14, SLX4, SPTA1, SPTB, STX11, STXB2, TBXA2R, TCIRG1, TERC, TERT, THBD, TINF2, TMPRSS6, TP53, TPI1, TUBB1, TYR, TYRP1, UNC13D, USB1, VKORC1, VWF, WAS, WRAP53, XIAP, XRCC2, YARS2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	ANEMIA	68	ABCB7, ADAMTS13, ALAS2, AMN, ANK1, ATM, ATR, ATRX, BLM, BRCA2, BRIP1, C15ORF41, CDAN1, CUBN, EPB42, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, G6PD, GATA1, GPI, GSS, HBA1, HBA2, HBB, HFE, KLF1, LPIN2, MTR, NBN, PALB2, PC, PDHA1, PDHX, PKLR, PUS1, RADS1C, RPL11, RPL15, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS29, RPS7, SBDS, SEC23B, SLC19A2, SLC4A1, SLX4, SPTA1, SPTB, THBD, TMPRSS6, TPI1, XRCC2, YARS2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	BLEEDING DISORDER/COAGULOPATHY	54	ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, AP3B1, BLOC1S3, BLOC1S6, CYCS, DTNBP1, F10, F11, F12, F13A1, F2, F5, F7, F8, F9, FGA, FGB, FGG, FLNA, GATA1, GGCX, GP1BA, GP1BB, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2B, ITGB3, LMNA1, MASTL, MPL, MYH9, NBEAL2, P2RY12, PROC, PROS1, RBM8A, RUNX1, SERPINC1, SLFN14, TBXA2R, TERC, TERT, TINF2, TP53, UNC13D, USB1, WAS, WRAP53, XIAP, XRCC2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	BLOOM SYNDROME	1	BLM	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	BONE MARROW FAILURE SYNDROME	93	ACTB, AK2, AP3B1, ATM, ATR, BLM, BLOC1S3, BLOC1S6, BRCA2, BRIP1, CDKN2A, CEBPA, CSF2RA, CTC1, CTSC, CXCR4, DKC1, DTNBP1, ELANE, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, G6PC3, GATA1, GATA2, HAX1, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, IFNGR2, ITK, JAGN1, KRAS, LYST, MAGT1, MLH1, MPL, MSH2, MSH6, MYO5A, NBN, NF1, NHP2, NOP10, NRAS, PALB2, PMS2, PRF1, PTPN11, RAB27A, RADS1C, RECQL4, RPL11, RPL15, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS29, RPS7, RTEL1, RUNX1, SBDS, SH2D1A, SLX4, STX11, STXB2, TCIRG1, TERC, TERT, TINF2, TP53, UNC13D, USB1, WAS, WRAP53, XIAP, XRCC2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	COAGULATION FACTOR DEFICIENCY	16	F10, F11, F12, F13A1, F2, F5, F7, F8, F9, FGA, FGB, FGG, GGCX, LMNA1, VKORC1, VWF	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	CONGENITAL NEUTROPENIA	13	ACTB, CSF2RA, CTSC, ELANE, G6PC3, GATA2, HAX1, IFNGR2, JAGN1, LYST, SBDS, TCIRG1, WAS	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	DIAMOND-BLACKFAN ANEMIA	12	GATA1, RPL11, RPL15, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS29, RPS7	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	DYSKERATOSIS CONGENITA	11	AK2, CTC1, DKC1, NHP2, NOP10, RTEL1, TERC, TERT, TINF2, USB1, WRAP53	Sequencing and Deletion-Duplication Testing of all Genes	1290

Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	FANCONI ANEMIA	22	ATM, ATR, BLM, BRCA2, BRIP1, CXCR4, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, NBN, PALB2, RAD51C, SLX4, XRCC2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS	13	FAS, ITK, LYST, MAGT1, MYO5A, PRF1, RAB27A, RECQL4, SH2D1A, STX11, STXBP2, UNC13D, XIAP	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	HEREDITARY LEUKEMIA	25	ATM, BLM, BRCA2, CDKN2A, CEBPA, DKC1, ELANE, FANCA, GATA2, HRAS, KRAS, MLH1, MSH2, MSH6, NBN, NF1, NRAS, PMS2, PTPN11, RUNX1, SBDS, TERC, TERT, TINF2, TP53	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	HERMANSKY-PUDLAK SYNDROME	22	ABCA3, AP3B1, BLOC1S3, BLOC1S6, DKC1, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, OCA2, SFTPB, SFTPC, SLC45A2, TERC, TERT, TINF2, TYR, TYRP1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	NEUTROPENIA	8	CSF3R, ELANE, G6PC3, GATA2, GFI1, HAX1, VPS45, WAS	Sequencing	1800
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	PLATELET FUNCTION DISORDER	18	AP3B1, BLOC1S3, BLOC1S6, DTNBP1, GP1BA, GP1BB, GP9, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA7B, ITGB3, NBEAL2, P2RY12, RUNX1, TRXA7R	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	RED BLOOD CELL MEMBRANE DISORDER	5	ANK1, EPB42, SLC4A1, SPTA1, SPTB	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	THROMBOCYTOPENIA	25	ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, CYCS, FLNA, GATA1, GP1BA, GP1BB, GP9, HOXA11, ITGA2, ITGA2B, ITGB3, MASTL, MPL, MYH9, NBEAL2, RBM8A, RUNX1, SLFN14, THBD, TUBB1, WAS	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	COMPREHENSIVE PRIMARY IMMUNODEFICIENCY	232	ACPS, ACTB, ADA, ADAR, ADIPOQ, ADIPOR1, ADIPOR2, AICDA, AIRE, AK2, AP3B1, ARMC4, ATM, BLM, BTK, C1QA, C1QB, C1QP, C1QC, C1R, C1S, C2, C3, C3AR1, C4A, C4B, C4BPA, C4BPB, C5, C5AR1, C5AR2, C6, C7, C8A, C8B, C8G, C9, CARD11, CARD14, CASP10, CASP8, CCDC103, CCDC114, CCDC39, CCDC40, CCDC65, CCNO, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD55, CD59, CD8A, CD93, CECR1, CFB, CFD, CFH, CFHR1, CFHR3, CFI, CFP, CIITA, CLU, COLEC11, CORO1A, CR1, CR2, CRP, CSF2RA, CTC1, CTLA4, CTSC, CYBA, CYBB, DCLRE1C, DDX58, DGKE, DKC1, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAAH1, DNAAH5, DNAAI1, DNAAI2, DNAL1, DNMT3B, DOCK2, DOCK8, DRC1, DYX1C1, ELANE, FAS, FCN1, FCN2, FCN3, FERMT3, FOXP3, G6PC3, G6PD, GATA2, HAX1, HYDIN, IFIH1, IFNGR1, IFNGR2, IGHM, IGLL1, IKBKG, IL10RA, IL10RB, IL12RB1, IL1RN, IL2RA, IL2RG, IL36RN, IL7, IL7R, ISG15, ITGB2, ITK, JAGN1, JAK3, LCK, LIG4, LPIN2, LRBA, LRRC6, LYST, MAGT1, MALT1, MASP1, MASP2, MAT2A, MEFV, MRE11A, MVK, NBN, NCF1, NCF2, NCF4, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRP12, NLRP3, NME8, NOD2, NOP10, NRAS, OFD1, ORAI1, PIGA, PIK3CD, PIK3R1, PLCG2, PMS2, PNP, PRF1, PRKDC, PSMB8, PSTPIP1, PTPRC, PTX3, RAB27A, RAG1, RAG2, RFX5, RFXANK, RFXAP, RHOH, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RPGR, RSPH1, RSPH4A, RSPH9, RTEL1, SAMHD1, SBDS, SERPING1, SH2D1A, SLC37A4, SMARCAL1, SP110, SPAG1, SPINK5, STAT1, STAT2, STAT3, STAT4, STAT5B, STIM1, STK4, STXBP2, TAP1, TAP2, TAPBP, TBX1, TCIRG1, TERC, TERT, THBD, TINF2, TMEM173, TNFRSF13B, TNFRSF1A, TNFRSF4, TRAC, TREX1, TYK2, UNC119, USB1, VSG14, VTN, WAS, WRAP53, XIAP, ZAP70, ZMYND10	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	AUTOINFLAMMATORY SYNDROME	25	ACPS, ADAR, CARD14, DDX58, ELANE, IFIH1, IL1RN, IL36RN, ISG15, LPIN2, MEFV, MVK, NLRP12, NLRP3, NOD2, PLCG2, PSMB8, PSTPIP1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TMEM173, TNFRSF1A, TREX1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	BONE MARROW FAILURE SYNDROME	93	ACTB, AK2, AP3B1, ATM, ATR, BLM, BLOC1S3, BLOC1S6, BRCA2, BRIP1, CDKN2A, CEBPA, CSF2RA, CTC1, CTSC, CXCR4, DKC1, DTNBP1, ELANE, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, G6PC3, GATA1, GATA2, HAX1, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, IFNGR2, ITK, JAGN1, KRAS, LYST, MAGT1, MLH1, MPL, MSH2, MSH6, MYO5A, NBN, NF1, NHP2, NOP10, NRAS, PALB2, PMS2, PRF1, PTPN11, RAB27A, RAD51C, RECQL4, RPL11, RPL15, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS29, RPS7, RTEL1, RUNX1, SBDS, SH2D1A, SLX4, STX11, STXBP2, TCIRG1, TERC, TERT, TINF2, TP53, UNC13D, USB1, WAS, WRAP53, XIAP, XRCC2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	CHRONIC GRANULOMATOUS DISEASE	7	CYBA, CYBB, G6PD, NCF1, NCF2, NCF4, NOD2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	COMPLEMENT SYSTEM DISORDER	82	ADIPOQ, ADIPOR1, ADIPOR2, ARMC4, C1QA, C1QB, C1QP, C1QC, C1R, C1S, C2, C3, C3AR1, C4A, C4B, C4BPA, C4BPB, C5, C5AR1, C5AR2, C6, C7, C8A, C8B, C8G, C9, CCDC103, CCDC114, CCDC39, CCDC40, CCDC65, CCNO, CD46, CD55, CD59, CD93, CFB, CFD, CFH, CFHR1, CFHR3, CFI, CFP, CLU, COLEC11, CR1, CR2, CRP, DGKE, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAAH1, DNAAH5, DNAAI1, DNAAI2, DNAL1, DRC1, DYX1C1, FCN1, FCN2, FCN3, HYDIN, LRRC6, MASP1, MASP2, MAT2A, NME8, OFD1, PIGA, PTX3, RPGR, RSPH1, RSPH4A, RSPH9, SERPING1, SPAG1, THBD, VSG14, VTN, ZMYND10	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	CONGENITAL NEUTROPENIA	13	ACTB, CSF2RA, CTSC, ELANE, G6PC3, GATA2, HAX1, IFNGR2, JAGN1, LYST, SBDS, TCIRG1, WAS	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	DYSKERATOSIS CONGENITA	11	AK2, CTC1, DKC1, NHP2, NOP10, RTEL1, TERC, TERT, TINF2, USB1, WRAP53	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	SEVERE COMBINED IMMUNODEFICIENCY	68	ADA, AK2, ATM, BLM, CARD11, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD8A, CIITA, CORO1A, DCLRE1C, DNMT3B, DOCK8, IFNGR1, IKBKG, IL12RB1, IL2RA, IL2RG, IL7R, ITGB2, ITK, JAK3, LCK, LIG4, LRBA, MAGT1, MALT1, NHEJ1, ORAI1, PIK3CD, PMS2, PNP, PRKDC, PTPRC, RAG1, RAG2, RFX5, RFXANK, RFXAP, RHOH, RMRP, RTEL1, SH2D1A, SMARCAL1, SP110, SPINK5, STAT1, STAT2, STAT3, STAT4, STAT5B, STIM1, STK4, TAP1, TAP2, TAPBP, TBX1, TNFRSF4, TRAC, TYK2, UNC119, WAS, ZAP70	Sequencing and Deletion-Duplication Testing of all Genes	1290

Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	COMPREHENSIVE CONGENITAL MALFORMATION AND SKELETAL DYSPLASIA	429	ABCC6, ACP5, ACTA1, ACTB, ACTG1, ACVR1, ADAMTS10, ADAMTS2, ADAMTSL2, ADGRG1, AGPS, AGRN, AKT1, AKT3, ALPL, ALX3, ALX4, AMER1, ANKH, ANOS5, ARFGEF2, ARHGAP31, ARID1A, ARID1B, ARSE, ARX, ASPA, ASPM, ASXL1, ATP6VOA2, ATR, B3GALNT2, B3GALT6, B4GALT7, BHLHA9, BIN1, BMP1, BMP2, BMP4, BMPR1A, BMPR1B, BRWD3, CA2, CANT1, CAPN3, CASK, CASR, CBS, CCM2, CCND2, CDC6, CDK5RAP2, CDKN1C, CDON, CDT1, CENPF, CENPJ, CEP152, CEP164, CEP63, CFL2, CHAT, CHD7, CHD8, CHKB, CHRNA1, CHRN1, CHRN2, CHRNE, CHRN3, CHST14, CHST3, CLCN5, CLCN7, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL4A2, COL4A4, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COL9A3, COLO, COMP, CREBBP, CRTAP, CSPP1, CTSK, CUL4B, CUL7, CYP27B1, DCX, DHCR24, DHCR7, DIS3L2, DLL3, DLL4, DLX3, DNM2, DNMT3A, DOCK6, DOK7, DPAGT1, DVL1, DYM, DYNC1H1, DYNC2H1, DYRK1A, EBP, ECEL1, EDN3, EDNRB, EFEMP2, EFN1, EFTUD2, EGR2, EIF2AK3, EIF2B5, ELN, EMD, EMX2, ENAM, ENPP1, EOGT, EP300, ERCC5, ERCC6, ERCC8, ESCO2, EVC, EVC2, EXOSC3, EXT1, EXT2, EYA1, EZH2, FAM20A, FAM20C, FAM83H, FANCB, FANCC, FBLN5, FBN1, FBN2, FGD1, FGF23, FGF3, FGF8, FGR1, FGR2, FGR3, FH, FHL1, FKBP10, FKBP14, FKRP, FKTN, FLNA, FLNB, FOXH1, FOXL2, FREM1, GAA, GBA, GBE1, GDF5, GFAP, GFPT1, GH1, GHR, GHRHR, GJA1, GLE1, GLI2, GLI3, GMPBP, GNAS, GNPAT, GPC3, GPSM2, GRIA3, HDAC8, HEPACAM, HESX1, HOXA13, HOXD13, HSPG2, HUWE1, IFT122, IFT140, IFT172, IFT80, IGF1, IGF1R, IGFALS, IHH, IKBKG, INSR, IRF6, IRS1, ISPD, KAT6B, KBTBD13, KDM6A, KIAA0196, KIF11, KIF1BP, KIF7, KIT, KLHL40, KMT2D, KRAS, KRIT1, L1CAM, LAMA2, LAMP2, LARGE, LBR, LEMD3, LHX3, LHX4, LIFR, LIG4, LMNA, LMX1B, LRP4, LRP5, LTBP2, MASP1, MATN3, MBD5, MCPH1, MED12, MEF2C, MID1, MITF, MLC1, MMP9, MPZ, MRE11A, MSX2, MTM1, MUSK, MYBPC1, MYH2, MYH3, MYH7, NALCN, NDE1, NEB, NEK1, NF1, NF2, NFIX, NHEJ1, NIPBL, NODAL, NOG, NOTCH1, NOTCH2, NPR2, NR5A1, NRG1, NSD1, NSDHL, OBSL1, OCLN, OFD1, OPHN1, ORC1, ORC4, ORC6, OTX2, P3H1, PAFAH1B1, PAPSS2, PAX3, PAX6, PCNT, PDCD10, PEX7, PHEX, PHF6, PIEZO2, PIGA, PIK3CA, PITX2, PLOD1, PLOD2, PMM2, PNKP, POLR1C, POMGNT1, POMT1, POMT2, POR, POU1F1, PP1B, PQBP1, PRKAR1A, PROX1, PTCH1, PTEN, PTH1R, PTPN11, PYCR1, RAB39B, RAB39A, RAB39B, RAB39A, RAB39B, RAF1, RAPS, RARS2, RASA1, RBBP8, RBP1, RECQL4, RELN, RET, RMRP, RNU4ATAC, ROR2, RUNX2, RYR1, SBDS, SCO2, SELENON, SERPINF1, SERPINH1, SF3B4, SKI, SOX10, TCF12, TGFBR1, TGFBR2, TTR, TWIST1, WDR19, WDR35	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	ACHONDROGENESIS, HYPOCHONDROGENESIS, FIBROCHONDROGENESIS	5	COL11A1, COL11A2, COL2A1, SLC26A2, TRIP11	Sequencing	1750
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	ADAMS-OLIVER SYNDROME	6	ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBP1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	ARTHROGYPOSES	58	ACTA1, AGRN, BIN1, CASK, CFL2, CHAT, CHRNA1, CHRN1, CHRN2, CHRNE, CHRN3, CHST14, COL6A2, COLO, DHCR24, DOK7, DPAGT1, ECEL1, EGR2, ERCC5, ERCC6, EXOSC3, FBN2, FHL1, FKTN, GBA, GBE1, GFPT1, GLE1, KAT6B, KLHL40, MPZ, MTM1, MUSK, MYBPC1, MYH2, MYH3, NALCN, NEB, PIEZO2, PLOD2, PMM2, RAPS, RARS2, SCO2, SELENON, TGFBR3, TK2, TNNI2, TNNT1, TPM2, TPM3, TRPV4, TSEN2, TSEN54, VPS33B,	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	BRACHYDACTYL / SYNDACTYL	12	BMPR1B, ESCO2, GDF5, GNAS, HOXA13, HOXD13, IHH, NOG, RECQL4, ROR2, SOX9, TP63	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	BRUCK SYNDROME	2	PLOD2, FKBP10	Sequencing	1000
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	CEREBRAL CAVERNOUS MALFORMATION	4	CCM2, KRIT1, PDCD10, RASA1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	CHONDRODYSPLASIA PUNCTATA	7	AGPS, ARSE, EBP, GNPAT, LBR, NSDHL, PEX7	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	COCKAYNE SYNDROME	2	ERCC6, ERCC8	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	CRANIOSYNOSTOSIS	34	ALPL, ALX3, ALX4, BMP4, EDN3, EDNRB, EFN1, ESCO2, FGF1, FGF2, FGF3, FLNB, FREM1, GDF5, GLI3, IFT122, IFT140, MASP1, MITF, MSX2, NOG, PAX3, POR, RECQL4, RET, SKI, SOX10, TCF12, TGFBR1, TGFBR2, TTR, TWIST1, WDR19, WDR35	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	DENTINOGENESIS IMPERFECTA, DENTIN DYSPLASIA, TOOTH AGENESIS	4	DSPP, MSX1, PTH1R, WNT10A	Sequencing	1750
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	DWARFISM	15	ATR, CDC6, CDT1, CENPJ, CEP152, CEP63, CUL7, NOTCH2, OBSL1, ORC1, ORC4, ORC6, PCNT, RBBP8, RNU4ATAC	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	ECTRODACTYL	4	LMBR1, TP63, WNT3, WNT10B	Sequencing	1750
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	FACIAL DYSOSTOSIS	29	ALPL, ALX3, ALX4, CREBBP, DLL3, EFN1, EFTUD2, EP300, EVC, EVC2, FGF1, FGF2, FGF3, GHR, HDAC8, HSPG2, LIFR, NIPBL, POLR1C, RMRP, SF3B4, SMC1A, SMC3, SOX9, SRCAP, TCF12, TCF1, TWIST1, UBE2A	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	FETAL AKINESIA	10	CHRNA1, CHRN2, CHRNE, CHRN3, CHST14, CHST3, CLCN5, CLCN7, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL4A2, COL4A4, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COL9A3, COLO, COMP, CREBBP, CRTAP, CSPP1, CTSK, CUL4B, CUL7, CYP27B1, DCX, DHCR24, DHCR7, DIS3L2, DLL3, DLL4, DLX3, DNM2, DNMT3A, DOCK6, DOK7, DPAGT1, DVL1, DYM, DYNC1H1, DYNC2H1, DYRK1A, EBP, ECEL1, EDN3, EDNRB, EFEMP2, EFN1, EFTUD2, EGR2, EIF2AK3, EIF2B5, ELN, EMD, EMX2, ENAM, ENPP1, EOGT, EP300, ERCC5, ERCC6, ERCC8, ESCO2, EVC, EVC2, EXOSC3, EXT1, EXT2, EYA1, EZH2, FAM20A, FAM20C, FAM83H, FANCB, FANCC, FBLN5, FBN1, FBN2, FGD1, FGF23, FGF3, FGF8, FGR1, FGR2, FGR3, FH, FHL1, FKBP10, FKBP14, FKRP, FKTN, FLNA, FLNB, FOXH1, FOXL2, FREM1, GAA, GBA, GBE1, GDF5, GFAP, GFPT1, GH1, GHR, GHRHR, GJA1, GLE1, GLI2, GLI3, GMPBP, GNAS, GNPAT, GPC3, GPSM2, GRIA3, HDAC8, HEPACAM, HESX1, HOXA13, HOXD13, HSPG2, HUWE1, IFT122, IFT140, IFT172, IFT80, IGF1, IGF1R, IGFALS, IHH, IKBKG, INSR, IRF6, IRS1, ISPD, KAT6B, KBTBD13, KDM6A, KIAA0196, KIF11, KIF1BP, KIF7, KIT, KLHL40, KMT2D, KRAS, KRIT1, L1CAM, LAMA2, LAMP2, LARGE, LBR, LEMD3, LHX3, LHX4, LIFR, LIG4, LMNA, LMX1B, LRP4, LRP5, LTBP2, MASP1, MATN3, MBD5, MCPH1, MED12, MEF2C, MID1, MITF, MLC1, MMP9, MPZ, MRE11A, MSX2, MTM1, MUSK, MYBPC1, MYH2, MYH3, MYH7, NALCN, NDE1, NEB, NEK1, NF1, NF2, NFIX, NHEJ1, NIPBL, NODAL, NOG, NOTCH1, NOTCH2, NPR2, NR5A1, NRG1, NSD1, NSDHL, OBSL1, OCLN, OFD1, OPHN1, ORC1, ORC4, ORC6, OTX2, P3H1, PAFAH1B1, PAPSS2, PAX3, PAX6, PCNT, PDCD10, PEX7, PHEX, PHF6, PIEZO2, PIGA, PIK3CA, PITX2, PLOD1, PLOD2, PMM2, PNKP, POLR1C, POMGNT1, POMT1, POMT2, POR, POU1F1, PP1B, PQBP1, PRKAR1A, PROX1, PTCH1, PTEN, PTH1R, PTPN11, PYCR1, RAB39B, RAB39A, RAB39B, RAB39A, RAB39B, RAF1, RAPS, RARS2, RASA1, RBBP8, RBP1, RECQL4, RELN, RET, RMRP, RNU4ATAC, ROR2, RUNX2, RYR1, SBDS, SCO2, SELENON, SERPINF1, SERPINH1, SF3B4, SKI, SOX10, TCF12, TGFBR1, TGFBR2, TTR, TWIST1, WDR19, WDR35	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	GASTROINTESTINAL ATRESIA	7	CHD7, FANCB, FANCC, GLI3, MID1, SOX2, TTC7A	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	HETEROTAXY / SITUS INVERSUS	25	ACVR2B, ANK56, CCDC105, CCDC11, CCDC114, CCDC39, CCDC40, DNAF1, DNAF2, DNAF3, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, FOXH1, GDF1, HEATR2, INVS, LEFTY2, LRRC6, NKX2	Sequencing	2100
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	HIRSCHSPRUNG DISEASE	9	EDN3, EDNRB, KIF1BP, MITF, NRG1, PAX3, RET, SOX10, ZEB2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	HOLOPROSENCEPHALY	12	CDON, FGF8, FGF1, FOXH1, GLI2, GLI3, NODAL, PTCH1, SHH, SIX3, TGIF1, ZIC2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	KABUKI SYNDROME	7	CHD7, EYA1, FLNB, IRF6, KDM6A, KMT2D, SIX5	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	LISSENCEPHALY	15	ACTB, ACTG1, ARX, DCX, FKTN, LARGE, PAFAH1B1, POMGNT1, POMT1, POMT2, RELN, TUBA1A, TUBB2B, VLDLR, YWHAE	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	MACROCEPHALY / OVERGROWTH SYNDROME	38	ABCC6, AKT1, AKT3, ASPA, BRWD3, CCND2, CDKN1C, CHD8, CUL4B, DHCR24, DIS3L2, DNMT3A, EIF2B5, EZH2, GFAP, GLI3, GPC3, GPSM2, GRIA3, HEPACAM, HUWE1, KIAA0196, KIF7, L1CAM, MED12, MLC1, NFIX, NSD1, OFD1, PIGA, PIK3CA, PTCH1, PTEN, RAB39B, RAB39A, RAB39B, RAF1, RAPS, RARS2, RASA1, RBBP8, RBP1, RECQL4, RELN, RET, RMRP, RNU4ATAC, ROR2, RUNX2, RYR1, SBDS, SCO2, SELENON, SERPINF1, SERPINH1, SF3B4, SKI, SOX10, TCF12, TGFBR1, TGFBR2, TTR, TWIST1, WDR19, WDR35	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	MEIER-GORLIN SYNDROME	5	CDC6, CDT1, ORC1, ORC4, ORC6	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	METAPHYSEAL DYSPLASIA	10	ANKH, CDKN1C, COL10A1, FGF3, FLNA, MMP9, PTH1R, RMRP, RUNX2, SBDS	Sequencing and Deletion-Duplication Testing of all Genes	1290

Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	MICROCEPHALY AND PONTOCEREBELLAR HYPOPLASIA	34	AKT3, ASPM, ATR, CASK, CDK5RAP2, CENPF, CENPI, CEP152, CEP164, CEP63, DYNC1H1, DYRK1A, EFTUD2, EXOSC3, KIF11, LIG4, MBD5, MCPH1, MRE11A, NDE1, NHE1, OPHN1, PAFAH1B1, PCNT, PNKP, POMT1, PQBP1, RARS2, STIL, TSEN2, TSEN54, TUBB2B, VRK1, WDR35	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	MICROMELIC DYSPLASIA	24	ADAMTS10, ADAMTS12, BMPR1B, DVL1, EXT1, FBN1, FGFR3, GDF5, GNAS, IFT122, IFT140, IHH, LIFR, LTBP2, NPR2, PRKAR1A, ROR2, SHOX, SMAD4, SOX9, TRPS1, WDR19, WDR35, WNT3A	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	MULTIPLE SYNSTOSES AND SYMPHALANGISM	6	FGF9, FLNB, GDF5, HOXA11, NOG, TTR	Sequencing	1750
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	NEUROFIBROMATOSIS	7	KIT, NF1, NF2, PTPN11, RAF1, SMARCB1, SPRED1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	NEURONAL MIGRATION DISORDER	47	ACTB, ACTG1, ADGRG1, AKT3, ARFGF2, ARX, B3GALNT2, CHD7, COL4A1, COL4A2, COL4A4, DCX, DYNC1H1, EMX2, FH, FKTN, FLNA, GMPBB, GPM2, ISPD, KIF1BP, KIF7, L1CAM, LAMA2, LARGE, MED12, MEF2C, NDE1, NSDHL, PAFAH1B1, PIK3CA, POMGNT1, POMT1, POMT2, RAB3GAP1, RAB3GAP2, RELN, SLC12A6, SRPX2, TMEM5, TUBA1A, TUBB2B, TUBB3, TUBB8, TUBG1, WDR62	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	OSTEOGENESIS IMPERFECTA	61	ACTA1, ALPL, ANOS, ATP6V0A2, B3GALNT2, B4GALT7, BMP1, CAPN3, CFL2, CHKB, CLCN5, COL1A1, COL1A2, COL6A1, COL6A2, COL6A3, CRTAP, DNM2, EMD, ENPP1, FGF23, FHL1, FKBP10, FKR, FKTN, FLNA, FLNB, GAA, GMPBB, ISPD, KBTBD13, KLHL40, LAMA2, LAMP2, LARGE, LMNA, LRP5, MYH7, NEB, OCRL, P3H1, PHEX, PIEZO2, PLOD2, POMGNT1, POMT1, POMT2, PPIB, PYCR1, RAPSN, RYR1, SELENON, SERPINF1, SERPINH1, SIL1, SLC34A3, TMEM43, TMEM5, TNNT1, TPM2, TPM3	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	OSTEOLYSIS	7	BANF1, MMP2, SOSTM1, TNFRSF11A, TREM2, TYROBP, WNK1	Sequencing	1750
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	OSTEOPETROSIS	21	AMER1, ANKH, CA2, CLCN7, COL1A1, CTSC, DLX3, FAM20C, GJA1, IKBKG, LEMD3, LRP4, LRP5, PTH1R, SLC29A3, SLC02A1, TCIRG1, TGFBI, TNFRSF11A, TNFRSF11B, TYROBP	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	OSTEOPETROSIS (PANEL 2)	6	CA2, CLCN7, OSTM1, PLEKHM1, TCIRG1, TNFRSF11A	Sequencing	1800
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	OSTEOPETROSIS (PANEL 3)	6	ANKH, DLX3, HPGD, LRP4, SOST, TGFBI, TNFRSF11B	Sequencing	1800
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	OSTEOPOROSIS	3	LRP5, WNT1, PLS3	Sequencing	1650
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	PATELLAR DYSOSTOSES	3	LMX1B, PITX1, TBX4	Sequencing	1850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	POLYDACTYLY - SYNDACTYLY - TRIPHALANGISM	6	GLI3, HOXD10, HOXD13, LMBR1, LRP4, PITX1	Sequencing	1850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	POLYMICROGYRIA	14	ADGRG1, AKT3, CHD7, FH, GPM2, KIF1BP, NDE1, NSDHL, SRPX2, TUBA1A, TUBA8, TUBB2B, TUBB3, WDR62	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	PSEUDOXANTHOMA ELASTICUM	4	ABCC6, ENPP1, GGCX, VEGFA (Hotspots)	Sequencing	1000
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	RHEUMATOID-LIKE OSTEOARTHROPATHIES	7	ACAN, COL2A1, HPGD, IL1RN, LPIN2, TRPV4, WISP3	Sequencing	1850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	SECKEL SYNDROME	6	ATR, CENPI, CEP152, CEP63, PCNT, RBBP8	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	SEPTO-OPTIC DYSPLASIA	4	HESX1, OTX2, PAX6, SOX2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	SEX DEVELOPMENT DISORDERS (DSD), HERMAPHRODITISM	29	AMH, AMHR2, AR, ARX, CBX2, CYP11A1, CYP11B1, CYP17A1, CYP19A1, DHCR7, DHH, DMRT1, HSD11B1, HSD17B3, HSD3B2, INSL3, LHCGR, MAMLD1, NR5A1, NR5A1, POR, RSPO1, RXRP2, SOX9, SRD5A2, STAR, TSPYL1, WT1, WNT4	Sequencing	1200
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	SHORT RIB DYSPLASIA / ASPHYXIATING THORACIC DYSPLASIA	15	CSPP1, DYNC2H1, EVC, EVC2, GLI2, IFT122, IFT140, IFT172, IFT80, NEK1, TCTN3, TTC21B, WDR19, WDR34, WDR35	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	SHORT STATURE	61	AKT1, ATR, BMP2, BMP4, BMPR1A, CDC6, CDT1, CENPI, CEP152, CEP63, CREBBP, CUL7, DHCR7, EP300, EYA1, FGD1, FGF3, FGFR3, FOXL2, GH1, GHR, GHRHR, GLI2, HESX1, IGF1, IGF1R, IGFALS, INSR, IRS1, KRAS, LHX3, LHX4, NIPBL, NOTCH2, NR5A1, OBSL1, ORC1, ORC4, ORC6, OTX2, PCNT, PITX2, POU1F1, PROP1, PTCH1, PTPN11, RAF1, RBBP8, RNU4ATAC, SHH, SHOX, SIX3, SMC1A, SOS1, SOX2, SOX3, STAT5B, TBX19, TBX3, TGIF1,	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	SKELETAL DYSPLASIA WITH ABNORMAL MINERALIZATION	27	ALPL, ANKH, B4GALT7, CASR, CLCN5, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, CRTAP, CYP27B1, ENPP1, FBN1, FGF23, FKBP10, P3H1, PHEX, PLOD2, PPIB, SERPINF1, SLC34A3, SLC39A13, SOX9, TNFRSF11A, TNFRSF11B, VDR	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	SPONDYLOMETAPHYSEAL / SPONDYLOEPI-(META)-PHYSEAL DYSPLASIA	18	ACP5, B3GALT6, CANT1, CHST3, COL11A1, COL11A2, COL2A1, DYM, EIF2AK3, HSPG2, MATN3, PAPSS2, RMRP, SLC39A13, SMARCA11, TRAPPC2, TRPV4, WISP3	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	STICKLER SYNDROME	8	COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, LRP2, VCAN	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	WARBURG MICRO SYNDROME	3	RAB3GAP1, RAB3GAP2, RAB18	Sequencing	3100

Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	COMPREHENSIVE METABOLISM	354	ABCC8, ABCD1, ABCD4, ACAA1, ACAD9, ACADL, ACADM, ACADS, ACADVL, ACAT1, ACOX1, ACSF3, ACY1, ADAMTSL2, ADAR, ADCK3, ADSL, AGA, AGK, AGL, AGPAT2, AKT2, ALAD, ALAS2, ALDH5A1, ALDH7A1, ALDOA, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, AMACR, AMT, ANO10, ANTXR2, APTX, ARG1, ARSA, ARSB, ASAH1, ASL, ASPA, ASS1, ATP13A2, ATP6VOA2, AUH, B3GLCT, B4GALT1, BCKDHA, BCKDHB, BSCL2, BTD, C10ORF2, C12ORF65, CACNA15, CAV1, CAV3, CBS, CD320, CKMT1A, CKMT1B, CKMT2, CLCN1, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CNMN1, CNNM2, CNNM4, COG1, COG4, COG5, COG6, COG7, COG8, COL11A2, COL2A1, COQ2, COQ6, COQ9, CPOX, CPS1, CPT1A, CPT1B, CPT2, CTNS, CTS, CTSC, CTSD, CTSK, DBT, DDOST, DGUOK, DHCR7, DHDDS, DLD, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DYM, ECHS1, EGF, ENO3, EPM2A, ETFA, ETFB, ETFDH, FBP1, FBXL4, FECH, FH, FLNA, FLNB, FOLR1, FUCA1, FXYP2, G6PC, GAA, GALT, GALNS, GAMT, GATM, GBA, GBE1, GCDH, GCH1, GCK, GIF, GLA, GLB1, GLDC, GLUD1, GMPPA, GNE, GNPTAB, GNPTG, GNS, GPC3, GPHN, GUSB, GYG1, GYS1, GYS2, HADH, HADHA, HADHB, HAMP, HCFC1, HEXA, HEXB, HFE, HFE2, HGSNAT, HLCS, HMBS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HPD, HRAS, HSD17B10, HSD17B4, HYAL1, IDS, IDUA, IFIH1, INSR, ISCU, IVD, KCNA1, KCNJ11, KCNJ2, L2HGDH, LAMA2, LAMP2, LDB3, LDHA, LIPA, LMBRD1, LMNA, LPIN1, MAGT1, MAN1B1, MANBA, MCCC1, MCCC2, MCEE, MCOLN1, MFN2, MFSDB, MGAT2, MMAA, MMB, MMACHC, MMADHC, MMGT1, MOCS1, MOCS2, MOGS, MPDU1, MPI, MPV17, MTHFR, MTR, MTRR, MUT, MYOT, NAGLU, NAGS, NDUFS1, NEU1, NHLRC1, NIP2A, NPC1, NPC2, OAT, OPA1, OPA3, OTC, OXCT1, PAH, PC, PCBD1, PCCA, PCCB, PCK1, PCK2, PDHA1, PDHB, PDHX, PDS1, PDS2, PDX1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX16, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG1, PHKG2, PHYH, PLIN1, PMM2, POLG, POLG2, PPARG, PPOX, PPT1, PRKAG2, PRKAG3, PRODH, PSAP, PTRF, PTS, PYGL, PYGM, QDPR, RAI1, RBCK1, RFT1, RNASEH2A, RNASEH2B, RNASEH2C, RPN2, RRM2B, RYR1, SAMHD1, SCN4A, SEC23B, S6SH, SLC12A3, SLC16A1, SLC17A5, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC25A3, SLC25A4, SLC2A2, SLC30A10, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A4, SLC3A1, SLC40A1, SLC41A2, SLC41A3, SLC46A1, SLC6A19, SLC6A8, SLC7A7, SLC7A9, SMPD1, SPG7, SRD5A3, SSR4, STT3A, STT3B, SUCLA2, SUCLG1, SUMF1, SUOX, TAZ, TBC1D4, TCF4, TCN2, TFR2, TFR3, TFR4, TFR5, TFR6, TFR7, TFR8, TFR9, TFR10, TFR11, TFR12, TFR13, TFR14, TFR15, TFR16, TFR17, TFR18, TFR19, TFR20, TFR21, TFR22, TFR23, TFR24, TFR25, TFR26, TFR27, TFR28, TFR29, TFR30, TFR31, TFR32, TFR33, TFR34, TFR35, TFR36, TFR37, TFR38, TFR39, TFR40, TFR41, TFR42, TFR43, TFR44, TFR45, TFR46, TFR47, TFR48, TFR49, TFR50, TFR51, TFR52, TFR53, TFR54, TFR55, TFR56, TFR57, TFR58, TFR59, TFR60, TFR61, TFR62, TFR63, TFR64, TFR65, TFR66, TFR67, TFR68, TFR69, TFR70, TFR71, TFR72, TFR73, TFR74, TFR75, TFR76, TFR77, TFR78, TFR79, TFR80, TFR81, TFR82, TFR83, TFR84, TFR85, TFR86, TFR87, TFR88, TFR89, TFR90, TFR91, 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TFR968, TFR969, TFR970, TFR971, TFR972, TFR973, TFR974, TFR975, TFR976, TFR977, TFR978, TFR979, TFR980, TFR981, TFR982, TFR983, TFR984, TFR985, TFR986, TFR987, TFR988, TFR989, TFR990, TFR991, TFR992, TFR993, TFR994, TFR995, TFR996, TFR997, TFR998, TFR999, TFR1000, TFR1001, TFR1002, TFR1003, TFR1004, TFR1005, TFR1006, TFR1007, TFR1008, TFR1009, TFR1010, TFR1011, TFR1012, TFR1013, TFR1014, TFR1015, TFR1016, TFR1017, TFR1018, TFR1019, TFR1020, TFR1021, TFR1022, TFR1023, TFR1024, TFR1025, TFR1026, TFR1027, TFR1028, TFR1029, TFR1030, TFR1031, TFR1032, TFR1033, TFR1034, TFR1035, TFR1036, TFR1037, TFR1038, TFR1039, TFR1040, TFR1041, TFR1042, TFR1043, TFR1044, TFR1045, TFR1046, TFR1047, TFR1048, TFR1049, TFR1050, TFR1051, TFR1052, TFR1053, TFR1054, TFR1055, TFR1056, TFR1057, TFR1058, TFR1059, TFR1060, TFR1061, TFR1062, TFR1063, TFR1064, TFR1065, TFR1066, TFR1067, TFR1068, TFR1069, TFR1070, TFR1071, TFR1072, TFR1073, TFR1074, TFR1075, TFR1076, TFR1077, TFR1078, TFR1079, TFR1080, TFR1081, 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Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	MITOCHONDRIAL DNA DEPLETION SYNDROME	25	AGK, APTX, AUH, C10ORF2, C12ORF65, DGUOK, FBXL4, MFN2, MPV17, NDUFS1, OPA1, OPA3, POLG, POLG2, RRM2B, SLC25A3, SLC25A4, SPG7, SUCLA2, SUCLG1, TIMM8A, TK2, TMEM126A, TYMP, WFS1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	MITONUCLEAR GENES	132	AARS2, ABCB7, ACAD9, ACADL, ACADM, ACADS, ACADVL, ADCK3, AFG3L2, AIFM1, ALAS2, APTX, ATP5E, ATPAF2, AUH, BCS1L, BOLA3, C10ORF2, C12ORF65, CISD2, COAS, COQ2, COQ6, COQ9, COX10, COX15, COX6B1, CPT1A, CPT2, DARS2, DGUOK, DLAT, DLD, DNAJC19, DNMI1, ETFA, ETFB, ETFDH, ETHE1, FASTKD2, FBP1, FH, FOXRED1, G6PC, GAMT, GATM, GFER, GFM1, GYS2, HARS2, HLCS, HADH, HADHA, HSPD1, ISCU, LRPPRC, MFN2, MPV17, MRPS16, MRPS22, MTFMT, MTPAP, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NUBPL, OPA1, OPA3, PC, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDX1, POLG, POLG2, PUS1, RARS2, REEP1, RRM2B, SARS2, SCO1, SCO2, SDHA, SDHAF1, SETX, SLC19A3, SLC25A20, SLC25A3, SLC25A4, SLC6A8, SLC37A4, SOD1, SPG7, SUCLA2, SUCLG1, SURF1, TACO1, TAZ, TIMM8A, TK2, TMEM126A, TMEM70, TRMU, TSFM, TTC19, TUFM, TYMP, UQCRRB, UQCRRQ	Sequencing	1800
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	OBESITY	36	ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, CRTC1, CUL4B, DYRK1B, GNAS, LEP, LEPR, MAGEL2, MCSR, MCR4, MKKS, MKS1, NROB2, NTRK2, PCSK1, PHF6, POMC, PPARG, PYY, SDCCAG8, SIM1, TRIM32, TTC8, UCP3, VPS13B, WDRCP	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	ORGANIC ACIDEMIA/ACIDURIA & COBALAMIN DEFICIENCY	32	ABCD4, ACAT1, ACSF3, BCKDHA, BCKDHB, CBS, CD320, DBT, DLD, ETFA, ETFB, ETFDH, GCDH, GIF, HCF1, HMGCL, IVD, LMBRD1, MCCCI, MCC2, MCEE, MMAA, MMAB, MMACHC, MMADHC, MTHFR, MTR, MTRR, MLIT, PCCA, PCCB, TCN1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	PERIODIC PARALYSIS	4	CACNA1S, CLCN1, KCNJ2, SCN4A	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	PEROXISOMAL DISORDERS	18	ABCD1, ACOX1, AMACR, GNPAT, HSD17B4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX16, PEX7, PEX7L, PEX3, PEX5, PEX6, PEX7, PHYH	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	PORPHYRIA	9	ALAD, ALAS2, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	ALPORT SYNDROME	6	CD151, COL4A3, COL4A4, COL4A5, COL4A6, MYH9	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	BARDET-BIEDL SYNDROME	18	ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, IFT172, MKKS, MKS1, SDCCAG8, TMEM67, TRIM32, TTC8	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	BARTTER SYNDROME	8	BSD, CASR, CLCNKA, CLCNKB, GNA11, KCNJ1, SLC12A1, SLC12A3	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	BRANCHIO-OTO-RENAL (BOR) SYNDROME	4	EYA1, SIX1, SIX5, TPAP2A	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	CILIOPATHY	83	AH1, ALMS1, ANKS6, ARL13B, ARL6, ARMC4, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C21ORF59, C5ORF42, CC2D2A, CCDC103, CCDC114, CCDC39, CCDC40, CCDC65, CCNO, CENPF, CEP164, CEP290, CEP41, CEP83, CFTR, CSPP1, DCDC2, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH1, DNAH5, DNAI1, DNAI2, DNAL1, DRC1, DYX1C1, GLIS2, HYDIN, IFT172, INPP5E, INVS, IQCB1, KIAA0586, KIF7, LRR6, MKKS, MKS1, NEK8, NME8, NPHP1, NPHP3, NPHP4, OFD1, RPGR, RPGRIP1L, RSPH1, RSPH4A, RSPH9, SDCCAG8, SPAG1, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TRIM32, TTC21B, TTC8, WDR19, ZMYND10, ZNF423	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	CYSTIC KIDNEY DISEASE	9	BICC1, EYA1, HNF1B, PAX2, PKD1, PKD2, PKHD1, SIX5, UMOD	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	DIABETES INSIPIDUS	3	AQP2, AVP, AVPR2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	HEMOLYTIC UREMIC SYNDROME	13	ADAMTS13, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, THBD	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	HYPOMAGNESEMIA	17	CLDN16, CLDN19, CNMN1, CNNM2, CNNM4, EGF, FXRD2, HNF1B, KCNA1, MAGT1, MMGT1, NIPA2, SLC12A3, SLC41A2, SLC41A3, TRPM6, TRPM7	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	HYPOPHOSPHATEMIC RICKETS	10	CLCN5, DMP1, ENPP1, FAH, FGF23, KL, PHEX, SLC34A1, SLC34A3, VDR	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	JOUBERT SYNDROME	29	AH1, ARL13B, B9D1, B9D2, C5ORF42, CC2D2A, CEP164, CEP290, CEP41, CSPP1, INPP5E, KIAA0586, KIF7, MKS1, NPHP1, NPHP3, OFD1, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	LIDDLE SYNDROME	2	SCNN1B, SCNN1G	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	MECKEL SYNDROME	11	B9D1, B9D2, CC2D2A, CEP290, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM216, TMEM231, TMEM67	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	MONOGENIC OBESITY	36	ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, CRTC1, CUL4B, DYRK1B, GNAS, LEP, LEPR, MAGEL2, MCSR, MCR4, MKKS, MKS1, NROB2, NTRK2, PCSK1, PHF6, POMC, PPARG, PYY, SDCCAG8, SIM1, TRIM32, TTC8, UCP3, VPS13B, WDRCP	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	NEPHRONOPHTHISIS	19	ANKS6, CEP164, CEP290, CEP83, DCDC2, GLIS2, IFT172, INVS, IQCB1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, TMEM67, TTC21B, WDR19, ZNF423	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	NEPHROTIC SYNDROME	29	ACTN4, ADCK4, ANLN, APOL1, ARHGAP24, ARHGDI1, CD2AP, COL4A3, COL4A4, COL4A5, COQ2, CRB2, DGKE, EMP2, INF2, ITGA3, LAMB2, LMX1B, MYH9, MYO1E, NPHS1, NPHS2, PICE1, PTPN8, SCARB2, SMARCA1, TRPC6, TTC21B, WT1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	POLYCYSTIC KIDNEY DISEASE	8	BICC1, LRPS, NOTCH2, PKD1, PKD2, PKHD1, PRKCSH, SEC63	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	PRIMARY CILIARY DYSKINESIA	32	ARMC4, C21ORF59, CCDC103, CCDC114, CCDC39, CCDC40, CCDC65, CCNO, CENPF, CFTR, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH1, DNAH5, DNAI1, DNAI2, DNAL1, DRC1, DYX1C1, HYDIN, INVS, LRR6, NME8, OFD1, RPGR, RSPH1, RSPH4A, RSPH9, SPAG1, ZMYND10	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	PRIMARY HYPEROXALURIA	3	AGXT, GRHPR, HOGA1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	PSEUDOHYPALDOSTERONISM	9	CUL3, HSD11B2, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	RENAL MALFORMATION	15	ACE, BMP4, DSTYK, EYA1, FANCB, FOXC2, FREM1, GATA3, HNF1B, PAX2, REN, RET, SIX1, SIX5, WT1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	RENAL TUBULAR ACIDOSIS	5	ATP6V0A4, ATP6V1B1, CA2, SLC4A1, SLC4A4	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	SENIOR-LOKEN SYNDROME	7	CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8	Sequencing and Deletion-Duplication Testing of all Genes	1290

Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	AMYOTROPHIC LATERAL SCLEROSIS (ALS)	31	ALS2, ANG, AT1L, BSCL2, CHCHD10, CHMP2B, DCTN1, FIG4, FUS, GBE1, GRN, HEXA, HNRNPA1, HSPD1, KIAA0196, KIF5A, OPTN, PRF1, REEP1, SETX, SLC52A2, SLC52A3, SOD1, SPAST, SPC11, SPC70, SOSTM1, TARDBP, UBQLN2, VAPB, VCP	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	ATAXIA	141	ABC7, ABHD12, ACO2, ADCK3, AFG3L2, AH1, ALDH5A1, ANO10, APTX, ARL13B, ARL6, ATCAY, ATM, ATN1, ATP8A2, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEAN1, C10ORF2, C5ORF42, CA8, CACNA1A, CACNB4, CAMTA1, CASK, CC2D2A, CCDC28B, CCDC88C, CEP290, CEP41, CLCN2, CLN5, CLPP, COX20, CSTB, CWF19L1, CYP27A1, DNAJC19, DNMT1, EEF2, ELOVL4, ELOVL5, FBXL4, FGF14, FLVCR1, FMR1, FXN, GBA2, GFAP, GOSR2, GRID2, GRM1, GSS, HARS2, HTT, INPP5E, ITM2B, ITPR1, KCNA1, KCNC3, KCND3, KCNU10, KIF1C, KIF7, LAMA1, LARS2, MARS2, MKKS, MKS1, MRE11A, MTPAP, MTPP, NEDD4, NOL3, NOP56, NPHP1, OFD1, OPA1, OPHN1, PAX6, PDYN, PEX7, PHYH, PNKD, PNKP, PNPLA6, POLG, PPP2R2B, PRKCG, PRRT2, RPRIP1L, RUBCN, SACS, SETX, SIL1, SLC1A3, SLC2A1, SLC52A2, SLC9A6, SNX14, SPG7, SPTBN2, STUB1, SYT14, TBP, TCTN1, TCTN2, TCTN3, TDP1, TGM6, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TPP1, TRIM32, TTBK2, TTC8, TTPA, TUBB4A, VAMP1, VLDLR, WDR45, WDR81, WFS1, WWOX, ZNF423, ZNF592	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	AUTISM SPECTRUM DISORDERS	13	CACNA1C, DHCRT, EN2, GAMT, MECP2, NLGN3, NLGN4X, NSD1, PDE8B, PTEN, RPL10, TSC1, TSC2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	CEROID LIPOFUSCINOSIS AND PROGRESSIVE MYOCLONIC EPILEPSY	28	AFG3L2, ASAH1, ATP13A2, CERS1, CLN3, CLN5, CLN6, CLN8, CSTB, CTSD, CTSF, DNAIC5, EPM2A, FOLR1, GOSR2, GRN, KCNC1, KCTD7, MFSDB, NEU1, NHLRC1, PPT1, PRICKLE1, PRICKLE2, SCARB2, SERPINI1, TRC1D24, TPP1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	CHARCOT-MARIE-TOOTH NEUROPATHY	86	AARS, AIFM1, AMACR, ARHGEF10, AT1L, AT1L3, ATP7A, BAG5, BSCL2, C12ORF65, CCT5, COX10, COX6A1, CTDP1, DCAF8, DCTN1, DHTKD1, DNMT2, DNMT1, DST, DYNC1H1, EGR2, FAM134B, FBLN5, FGD4, FIG4, FXN, GAN, GARS, GDAP1, GJB1, GNB4, GNE, HADHB, HARS, HINT1, HK1, HSPB1, HSPB8, IGHMBP2, INF2, KARS, KIF1A, KIF1B, KIF5A, LDB3, LITAF, LMNA, LRSAM1, MARS, MED25, MFN2, MPZ, MTRMR2, MYOT, NDRG1, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP22, POLG, PRPS1, PRX, RAB7A, REEP1, SACS, SBF1, SBF2, SCN9A, SETX, SH3TC2, SLC12A6, SMAD3, SPG11, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TYMP, VCP, WNK1, YARS	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	COENZYME Q10 DEFICIENCY	11	ADCK3, ANO10, APTX, COO2, COQ6, COQ9, ETFA, ETFB, ETFDH, PDSS1, PDSS2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	CONGENITAL HYPOVENTILATION	7	ASCL1, BDNF, BMP2, EDN3, PHOX2A, PHOX2B, RET	Sequencing	1650
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MYASTHENIA	17	AGRN, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COLQ, DOK7, DPAGT1, GFPT1, LAMB2, MIISK, PLEC, RAPSIN, SCN4A, STIM1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	DEMENTIA	17	APOE, APP, CHMP2B, CSF1R, FUS, GRN, MAPT, PRNP, PSEN1, PSEN2, SIGMAR1, SORL1, TARDBP, TREM2, UBE3A, UBQLN2, VCP	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	DYSTONIA	6	GCH1 (DYT5), TH, SGCE (DYT11), SPR, ATP1A3 (DYT12), PRKRA (DYT16)	Sequencing	2900
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	EPILEPSY	194	ABCD1, ADAR, ADSL, AFG3L2, AGA, AIMP1, ALDH5A1, ALDH7A1, ALG13, AMACR, AMT, ARG1, ARHGEF9, ARSA, ARX, ASAH1, ASPA, ATP13A2, ATRX, BTBD, CACNA1A, CACNA1H, CACNB4, CASK, CASR, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN2, CLN3, CLN5, CLN6, CLN8, CNTNAP2, COL4A1, COX15, CPT2, CSF1R, CSTB, CTSD, CTSF, CUL4B, DARS2, DCX, DEPDC5, DNAIC5, DNMT1, DOCK7, DPYD, EARS2, EEF1A2, EFHC1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPM2A, ETFA, ETFB, ETFDH, FAM126A, FH, FLNA, FOLR1, FOXG1, FOXRED1, GABRA1, GABRB3, GABRG2, GALT, GAMT, GCDH, GCH1, GFAP, GJC2, GLDC, GNAO1, GNE, GOSR2, GPHN, GRIA3, GRIN2A, GRIN2B, GRN, HCN1, HEPACAM, HNRNPU, HSD17B10, HSPD1, IQSEC2, KCNA1, KCNA2, KCNB1, KCNC1, KCNQ2, KCNQ3, KCNT1, KCTD7, KDM5C, KIF1A, L2HGDH, LGI1, MARS2, MBDS, MECP2, MED12, MEF2C, MFSDB, MLC1, MOCS1, MTHFR, MTOR, NDUFAF5, NECAP1, NEU1, NHLRC1, NOTCH3, NRXN1, OFD1, OPHN1, PCDH19, PGK1, PHF6, PIGA, PLCB1, PLP1, PNKP, PNPO, POLR3A, POLR3B, PPT1, PRICKLE1, PRICKLE2, PRODH, PRRT2, PSAP, PTS, PURA, QDPR, RAB39B, RELN, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SERPINI1, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC25A15, SLC25A22, SLC2A1, SLC35A2, SLC46A1, SLC6A1, SLC6A8, SLC9A6, SMS, SNAP25, SOX10, SPTAN1, ST3GAL3, ST3GAL5, STX1B, STXBP1, SUMF1, SUOX, SYN1, SYNGAP1, SZT2, TBC1D24, TCF4, TPP1, TREX1, TSC1, TSC2, TUBB4A, UBE2A, UBE3A, WDR45, WWOX, ZEB2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	EPILEPSY AND X-LINKED DEVELOPMENTAL DELAY	22	ARHGEF9, ARX, ATRX, CASK, CDKL5, CUL4B, GRIA3, HSD17B10, IQSEC2, KDM5C, MECP2, MED12, OFD1, OPHN1, PCDH19, PGK1, PHF6, RAB39B, SLC9A6, SMS, SYN1, UBE2A		
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	EPILEPTIC ENCEPHALOPATHY	83	ADAR, ADSL, ALDH7A1, ALG13, AMT, ARHGEF9, ARX, CACNA1A, CASK, CDKL5, CHD2, CNTNAP2, CPT2, DCX, DNMT1, DOCK7, EEF1A2, FLNA, FOXG1, GABRA1, GABRB3, GABRG2, GAMT, GLDC, GNAO1, GPHN, GRIN2A, GRIN2B, HCN1, HEPACAM, HNRNPU, KCNA2, KCNB1, KCNQ2, KCNQ3, KCNT1, KIF1A, MBDS, MECP2, MEF2C, MOCS1, MTHFR, NECAP1, NRXN1, PCDH19, PIGA, PLCB1, PNKP, PNPO, PURA, RNASEH2A, RNASEH2B, SAMHD1, SCN1A, SCN1B, SCN2A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC25A22, SLC2A1, SLC35A2, SLC6A8, SLC9A6, SNAP25, SPTAN1, ST3GAL3, ST3GAL5, STXBP1, SYN1, SYNGAP1, SZT2, TBC1D24, TCF4, TREX1, TSC1, TSC2, UBE3A, WDR45, WWOX, ZEB2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	LEUKODYSTROPHY AND LEUKOENCEPHALOPATHY	42	ABCD1, ADAR, AIMP1, ARSA, ASPA, CLCN2, COL4A1, COX15, CSF1R, DARS2, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, FAM126A, FOLR1, FOXRED1, GALT, GFAP, GJC2, HEPACAM, HSPD1, L2HGDH, MARS2, MLC1, NDUFAF5, NOTCH3, PLP1, POLR3A, POLR3B, PSAP, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SOX10, SUMF1, TREX1, WDR45	Sequencing and Deletion-Duplication Testing of all Genes	1290

Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MENTAL RETARDATION (AUTOSOMAL RECESSIVE)	20	AP4B1, AP4E1, AP4M1, AP4S1, CA8, CC2D1A, CNTNAP2, CRBN, ERLIN2, GRIK2, MAN1B1, NRXN1, PRSS12, ST3GAL3, STXBP1, TRAPPC9, TUSC3, VLDLR, ZC3H14, ZNF526	Sequencing	3950
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MENTAL RETARDATION (X-LINKED)	94	ABCD1, ACSL4, AFF2, AGR2, AP152, ARHGGEF6, ARHGGEF9, ARX, ATP6A2, ATP7A, ATRX, BCOR, BRWD3, CASK, CDKL5, CUL4B, DCX, DKC1, DLG3, ELK1, FANCB, FGD1, FLNA, FMR1, FTS1, GDI1, GK, GPC3, GRIA3, HCCS, HPRT1, HSD17B10, HUWE1, IDS, IGBP1, IL1RAPL1, IQSEC2, KDM5C, KIAA2022, KLF8, L1CAM, LAMP2, MAGT1, MAOA, MBTPS2, MECP2, MED12, MID1, MTM1, NDP, NDUFA1, NHS, NLGN3, NLGN4X, NSDHL, NXF5, OCRL, OFD1, OPHN1, OTC, PAK3, PCDH19, PDHA1, PGK1, PHF6, PHF8, PLP1, PORCN, PQBP1, PRPS1, RAB39B, RPL10, RPS6KA3, SHROOM4, SLC16A2, SLC6A8, SLC9A6, SMC1A, SMS, SOX3, SRPX2, SYN1, SYP, TIMM8A, TSPAN7, UBE2A, UPP3B, ZCCHC12, ZDHC15, ZDHC9, ZNF41, ZNF674, ZNF711, ZNF81	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MICROCEPHALY	24	AP4M1, ASPM, CASC5, CASK, CDK5RAP2, CENPJ, CEP63, CEP135, CEP152, EFTUD2, IER3IP1, KIF11, MCPH1, NDE1, NHEJ1, PAFAH1B1, PCNT, PNKP, POMT1, SLC25A19, STIL, TUBB2B, TUBGCP6, WDR62	Sequencing	1800
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MIGRAINE	10	ATP1A2, ATP1A3, CACNA1A, KCNK18, NOTCH3, POLG, PRRT2, SCN1A, SLC1A3, SLC2A1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MUSCULAR DYSTROPHY (COLLAGEN TYPE VI-RELATED)	6	COL12A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MUSCULAR DYSTROPHY (CONGENITAL)	25	B3GALNT2, B3GNT1, CHKB, COL12A1, COL6A1, COL6A3, DAG1, DPM1, DPM3, FKR, FKTN, GMPPB, GOSR2, ISPD, ITGA7, LAMA2, LARGE, LMNA, POMGNT1, POMGNT2, POMK, POMT1, POMT2, ST3GAL4, TMEM5	Sequencing	2100
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MUSCULAR DYSTROPHY (EMERY-DREIFUSS)	6	DMD, EMD, FHL1, LMNA, TMEM43, TTN	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MUSCULAR DYSTROPHY (LIMB GIRDLE AND CONGENITAL)	33	ANOS, CAPN3, CAV3, COL4A1, COL4A2, DES, DMD, DNAJB6, DYSF, FKR, FKTN, GMPPB, ISPD, LAMA2, LARGE, LIMS2, LMNA, MYOT, PNPLA2, POMGNT1, POMT1, POMT2, SGCA, SGCB, SGCD, SGGC, SMCHD1, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MUSCULAR DYSTROPHY / MYOPATHY	51	ACTA1, ANOS, CAPN3, CAV3, CFL2, COL12A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, DES, DMD, DNAJB6, DYSF, EMD, FHL1, FKR, FKTN, GMPPB, ISPD, KBTBD13, KLHL40, KLHL41, LAMA2, LARGE, LIMS2, LMNA, LMOD3, MTM1, MYOT, NEB, PNPLA2, POMGNT1, POMT1, POMT2, SGCA, SGCB, SGCD, SGGC, SMCHD1, TCAP, TMEM43, TNNT1, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TTN	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MYASTHENIA	17	AGRN, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COLQ, DOK7, DPAGT1, GFPT1, LAMB2, MANK, PLEC, RAPS, SCN4A, STIM1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MYASTHENIA (PANEL 2)	40	AGRN, BLK, C5, CCL21, CCR6, CD2, CD28, CD40, CD58, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COLQ, DOK7, GFPT1, IL2, IL21, IL2RA, IL2RB, IL6ST, IRF5, MUSK, PRDM1, PRKCO, PTPRC, RAG1, RAPS, RBPJ, SCN4A, STAT4, TAGAP, TNFAIP3, TNFRSF14, TRAF1, TRAF2, TRAF3, TRAF6, VAMP2	Sequencing	2050
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MYOPATHY	34	ACTA1, ATP2A1, BAG3, BIN1, CAV3, CFL2, CLCN1, CNTN1, COL6A1, COL6A2, COL6A3, CRYAB, DES, DNM2, DYSF, FHL1, FLNC, GNE, ISCU, KBTBD13, LDB3, MATR3, MTM1, MYH2, MYH7, MYOT, NEB, RYR1, SEPN1, TNNT1, TPM2, TPM3, TTN, VCP	Sequencing	2050
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MYOPATHY (CENTRONUCLEAR)	6	BIN1, CCDC78, DNM2, MTM1, MYF6, RYR1	Sequencing	1700
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MYOPATHY (DISTAL)	18	ANOS, BAG3, CAV3, CRYAB, DES, DNAJB6, DYSF, FHL1, FLNC, GNE, LDB3, MATR3, MYH7, MYOT, TCAP, TIA1, TTN, VCP	Sequencing	1800
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MYOPATHY (NEMALINE)	11	ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, MTM1, NEB, TNNT1, TPM2, TPM3		
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	NEURODEGENERATION WITH BRAIN IRON ACCUMULATION (NRIA)	7	PANK2, PLA2G6, C19ORF12, FTL, FA2H, ATP13A2, CP	Sequencing	2950
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	PARKINSON DISEASE	12	ATP13A2, DNAJC6, FBXO7, LRRK2, MAPT, PARK2, PARK7, PINK1, PLA2G6, SLC6A3, SNCA, VPS35	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	PARKINSON DISEASE (PANEL 2)	23	ADH1C, ATP13A2, ATP1A3, DCTN1, EIF4G1, FBXO7, GBA, GCH1, GIGYF2, HTRA2, LRRK2, MAPT, PARK2, PARK7, PDXK, PINK1, PLA2G6, POLG1, SNCA, SNCAIP, SNCB, UCHL1, VPS35	Sequencing	2050
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	PARKINSON DISEASE WITH DYSTONIA	5	TAH1, SLC6A3, ATP1A3, PRKRA, PLA2G6	Sequencing	2750
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	PERIODIC PARALYSIS	4	CACNA1S, CLCN1, KCNJ2, SCN4A	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	SPASTIC PARAPLEGIA	35	AFG3L2, ALS2, AT1L1, BAGALNT1, BSCL2, C12ORF65, C19ORF12, CYP7B1, DDHD1, DDHD2, FA2H, FXN, GALC, GBA2, GIC2, HSPD1, KDM5C, KIAA0196, KIF1A, KIF5A, L1CAM, MARS2, NIPA1, PLP1, PNPLA6, REEP1, SACS, SETX, SLC16A2, SLC33A1, SPAST, SPG11, SPG20, SPG7, SPG8	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	SPASTIC PARAPLEGIA (PANEL 2)	27	ALS2, AP5Z1, AT1L1, BSCL2, CCT5, CYP7B1, FA2H, GIC2, HSPD1, KDM5C, KIAA0196, KIF1A, KIF5A, L1CAM, NIPA1, PLP1, PNPLA6, REEP1, SLC16A2, SLC33A1, SPAST, SPG7, SPG11, SPG20, SPG7, SPG8, ZFYVE26, ZFYVE27	Sequencing	2100
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	SPINAL MUSCULAR ATROPHY	29	AARS, ASAH1, ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, EXOSC3, EXOSC8, FBXO38, GARS, HEXA, HSPB1, HSPB3, HSPB8, IGHMBP2, LAS1L, PLEKHG5, REEP1, SCO2, SLC5A7, SMN1, SMN2, TRPV4, UBR1, VAPB, VPRK1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	ZELLWEGER SYNDROME	14	EX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHYH	Sequencing	1500
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	ACHROMATOPSIA	6	ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	ALBINISM	19	AP3B1, BLOC1S3, BLOC1S6, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, MC1R, MITE, MYO5A, OCA2, RAB27A, SLC45A2, TYR, TYRP1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	BARDET-BIEDL SYNDROME	18	ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, IFT172, MKKS, MKS1, SDC CAG8, TMEM67, TRIM32, TTC8	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	BRITTLE CORNEA	2		Sequencing	600

Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	CATARACT	54	ABCB6, ADAMTSL4, AGK, ALDH18A1, BCOR, BFSP2, COL11A1, COL18A1, COL2A1, COL4A1, CRYAA, CRYAB, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CYP27A1, ERCC2, ERCC5, ERCC6, ERCC8, EYA1, FAM126A, FOXE3, FTL, FYCO1, FZD4, GALK1, GALT, GCNT2, GJA1, GJA3, GJA8, HSF4, LIM2, MAF, MYH9, NDP, NF2, NHS, OCRL, OPA3, PAX6, PITX3, RAB3GAP1, REQL4, SIL1, SLC33A1, TDRD7, TFAP2A, TMEM70, WFS1, WRN	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	CONE ROD DYSTROPHY	32	ABCA4, ADAM9, AIPL1, BEST1, C8ORF37, CABP4, CACNA1F, CACNA2D4, CDHR1, CERKL, CLN3, CNGA3, CNGB3, CNMN4, CRB1, CRX, CYP4V2, FBLN5, GNAT2, GUCA1A, GUCY2D, KCNV2, MERK, PDE6C, PDE6H, PROM1, PRPH2, RAX2, RDH5, RPGR, RPRIP1, SEMA4A	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	CONGENITAL STATIONARY NIGHT BLINDNESS	17	CABP4, CACNA1F, CACNA2D4, CYP4V2, GNAT1, GPR179, GRK1, GRM6, LRIT3, NYX, PDE6B, RDH5, RHO, RLBP1, RPE65, SAG, TRPM1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	CORNEAL DYSTROPHY	15	CHST6, COL5A1, CYP4V2, FOXE3, GJA8, KRT12, LCAT, LOXHD1, MAF, PITX2, SLC4A11, TCF4, TGFB1, ZNF469	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	CORNEAL DYSTROPHY (PANEL 2)	20	CHST6, COL5A1, COL8A2, CYP4V2, DCN, GSN, KRT3, KRT12, LOXHD1, PIKFYVE, PRDM5, SLC4A11, SOD1, ZNF1, ZNF469, TACSTD2, TCF4, TGFB1, IIRIAD1, V5X1	Sequencing	1550
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	ECTOPIA LENTIS	3	LTBP2, ADAMTSL4, FBN1	Sequencing	1600
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	FLECKED RETINA DISORDERS	11	ABCA4, CHM, CYP4V2, ELOVL4, PROM1, PRPH2, RDH5, RHO, RLBP1, RS1, VPS13F	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	GLAUCOMA	17	CNTNAP2, COL4A1, CYP1B1, FOXC1, FOXE3, LMX1B, LTBP2, MAF, MYOC, OPA1, OPA3, OPTN, PAX6, PITX2, TRK1, TMEM126A, WDR36	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	JOUBERT SYNDROME	29	AHI1, ARL13B, B9D1, B9D2, C5ORF42, C2D2A, CEP164, CEP290, CEP41, CSPP1, INPP5E, KIAA0586, KIF7, MKS1, NPHP1, NPHP3, OFD1, RPRIP1, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	LEBER CONGENITAL AMAUROSIS	25	AIPL1, ALMS1, BBS4, CABP4, CEP290, CNGA3, CRB1, CRX, DTHD1, GUCY2D, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, MERK, MYO7A, NMNAT1, RD3, RDH12, RDH5, RPE65, RPRIP1, SPATA7, TULP1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	MACULAR DYSTROPHY	17	ABCA4, BEST1, CERKL, CNGB3, CRB1, ELOVL4, FBLN5, IMPG1, PROM1, PRPH2, RAX2, RDH12, RDH5, RLBP1, RPL11, RPRG, RS1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	MACULAR DYSTROPHY (PANEL 2)	21	ABCA4, ARMS2, C2, C3, C9, CCR3, CFB, CFH, CFI, CST3, CXCL8, CX3CR1, ERCC6, FBLN5, HMCN1, HTRA1, IL6, IL1A, NLRP3, RAX2, TLR4	Sequencing	1850
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	MICROPHthalmia, ANOPHTHALMIA AND ANTERIOR SEGMENT DYSGENESIS	32	ABCB6, BCOR, BMP4, CHD7, COL4A1, CYP1B1, ERCC2, ERCC5, ERCC6, FOXC1, FOXE3, FOXL2, FRAS1, FREM1, GJA1, HCCS, HESX1, NDP, OCRL, OTX2, PAX2, PAX6, PITX2, PQBP1, RAB3GAP1, SHH, SIX3, SOX2, STRA6, TFAP2A, VPS13B, ZIC2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	NEURO-OPHTHALMOLOGY	26	APTX, C10ORF2, C12ORF65, FRMD7, GPR143, HESX1, MFN2, NDUFS1, OPA1, OPA3, OTX2, PAX6, POLG, ROBO3, RRM2B, SALL4, SETX, SLC25A4, SOX2, SPG7, TIMM8A, TK2, TMEM126A, TUBB3, TYMP, WFS1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	OPTIC ATROPHY	10	C12ORF65, MFN2, NDUFS1, OPA1, OPA3, POLG, SPG7, TIMM8A, TMEM126A, WFS1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	RETINAL DYSTROPHY	181	ABCA4, ABHD12, ADAM9, ADGRV1, AHI1, AIPL1, ALMS1, ARL13B, ARL6, ATF6, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C2ORF71, C5ORF42, C8ORF37, CABP4, CACNA1F, CACNA2D4, CAPN5, C2D2A, CDH23, CDHR1, CEP164, CEP290, CEP41, CERKL, CHM, CIB2, CLN3, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNMN4, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, CRB1, CRX, CSPP1, CYP4V2, DFNB31, DHDD5, DTHD1, EFEMP1, ELOVL4, EYS, FAM161A, FBLN5, FLVCR1, FRMD7, FZD4, GNAT1, GNAT2, GNPTG, GPR179, GRK1, GRM6, GUCA1A, GUCY2D, HARS, HK1, HMX1, IDH3B, IFT140, IFT172, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, KCNJ13, KCNV2, KIAA0586, KIF11, KIF7, KLHL7, LCA5, LRAT, LRIT3, LRP2, LRP5, MAK, MERK, MKKS, MKS1, MVK, MYO7A, NDP, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, NYX, OAT, OFD1, OPA1, OPA3, OTX2, PANK2, PCDH15, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PDZD7, PEX1, PEX2, PEX7, PHYH, PRCD, PROM1, PRPF3, PRPF51, PRPF8, PRPH2, RAX2, RBP3, RD3, RDH12, RDH5, RGR, RHO, RLBP1, RPL11, RPL2, RPE65, RPGR, RPRIP1, RPRIP1L, RS1, SAG, SDCCAG8, SEMA4A, SNRNP200, SPATA7, TCTN1, TCTN2, TCTN3, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TRIM32, TRPM1, TSPAN12, TTC21B, TTC8, TTPA, TULP1, USH1C, USH1G, USH2A, VCAN, VPS13B, WDR19, ZNF423, ZNF513	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	RETINITIS PIGMENTOSA	80	ABCA4, ABHD12, AIPL1, ARL6, BBS1, BBS2, BEST1, C2ORF71, C8ORF37, CDHR1, CEP290, CERKL, CHM, CLN3, CLRN1, CNGA1, CNGB1, CRB1, CRX, CYP4V2, DHDD5, EYS, FAM161A, FLVCR1, GNPTG, GUCY2D, HK1, IDH3B, IMPDH1, IMPG2, KLHL7, LCA5, LRAT, MAK, MERK, MVK, NMNAT1, NR2E3, NRL, OAT, OFD1, PANK2, PDE6A, PDE6B, PDE6G, PEX1, PEX2, PEX7, PHYH, PRCD, PROM1, PRPF3, PRPF51, PRPF8, PRPH2, RBP3, RDH12, RDH5, RGR, RHO, RLBP1, RPL1, RPL2, RPE65, RPGR, RPRIP1, RS1, SAG, SEMA4A, SNRNP200, SPATA7, TOPORS, TTC8, TTPA, TULP1, USH1C, USH2A, VPS13B, WDR19, ZNF513	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	RETINITIS PIGMENTOSA (AUTOSOMAL DOMINANT) (PANEL 3)	21	ASCC3L1, CM, CRX, FSCN2, GUCA1B, IMPDH1, KU-IL7, NR2E3, NRL, PRPF3, PRPF8, PRPF31, PRPH2-RDS, RDH12, RHO, ROM1, RP1, RP9, SEMA4A, TOPORS, VMD2-BEST1	Sequencing	2600
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	RETINITIS PIGMENTOSA (AUTOSOMAL RECESSIVE) (PANEL 2)	56	BCA4, ASCC3L1, BEST1, C2ORF71, C8ORF37, CA4, CERKL, CLRN1, CNGA1, CNGB1, CRB1, CRX, DHDD5, EYS, FAM161A, FSCN2, GUCA1B, IDH3B, IMPDH1, IMPG2, KLHL7, LRAT, MAK, MERK, NR2E3, NRL, OFD, PDE6A, PDE6B, PDE6G, PRCD, PROM1, PRPF3, PRPF6, PRPF8, PRPF31, PRPH2, RBP3, RDH12, RGR, RHO, RLBP1, ROM1, RP1, RP2, RP9, RPE65, RPGR, SAG, SEMA4A, SPATA7, TTC8, TOPORS, TULP1, USH2A, ZNF513	Sequencing	1600
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	RETINOBLASTOMA	1	RB1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	SENIOR-LOKEN SYNDROME	7	CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8	Sequencing and Deletion-Duplication Testing of all Genes	1290

Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	STARGARDT DISEASE AND MACULAR DYSTROPHIES	13	ABCA4, BEST1, C10TNF5, CDH3, CNGB3, ELOVL4, FSCN2, PROM1, PRPH2, RDH12, RPL11, RPGR, TIMP3	Sequencing	2950
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	STICKLER SYNDROME	8	COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, LRP2, VCAN	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	USHER SYNDROME	13	ABHD12, ADGRV1, CDH23, CIB2, CLRN1, DFNB31, HARS, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	USHER SYNDROME (PANEL 2)	12	ABHD12, CDH23, CLRN1, DFNB31, GPR98, HARS/USH3B, MYO7A, PCDH15, USH1C, USH1G, USH1I, USH2A	Sequencing	2100
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	USHER SYNDROME (PANEL 3)	20	ABHD12, CDH23, CIB2, CLRN1, COL4A6, DFNB31, DSPP (Excluding Exon 5), GIPC3, GPR98, HARS, KARS, LHFPL5, LOXHD1, MYO7A, PCDH15, PDZD7, TNC, USH2A, USH1C, USH1G	Sequencing	1490
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	VITREORETINOPATHY	17	BEST1, CAPN5, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, FZD4, KCNN13, LRP5, NDP, NR2F3, BS1, TSPAN12, VCAN	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	COMPREHENSIVE PULMONOLOGY	61	ABCA3, CCDC39, CCDC40, CFTR, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, CSF2RA, DKC1, DAAAF1, DAAAF2, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, EDN3, EFEMP2, ELMOD2, ELN, FBLN5, FLCN, FOXF1, GLRA1, HPS1, HPS4, ITGA3, LTBP4, MECP2, NF1, NKX2-1, NME8, PARN, PHOX2B, RAPSN, RET, RSPH4A, RSPH9, RTEL1, SCN4A, SCNN1A, SCNN1B, SERPINA1, SFTPA1, SFTPA2, SFTPB, SFTPC, SLC34A2, SLC6A5, SLC7A7, SMPD1, STAT3, TERC, TERT, TINF2, TSC1, TSC2, ZEB2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	BRONCHIECTASIS	15	CCDC39, CCDC40, CFTR, DAAAF1, DAAAF2, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, NME8, RSPH4A, RSPH9, SCNN1A, SCNN1B	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	CENTRAL HYPOVENTILATION AND APNEA	15	CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, EDN3, GLRA1, MECP2, PHOX2B, RAPSN, RET, SCN4A, SLC6A5, ZEB2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	CYSTIC LUNG DISEASE	8	EFEMP2, ELN, FBLN5, FLCN, LTBP4, SERPINA1, TSC1, TSC2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	HERMANSKY-PUDLAK SYNDROME	22	ABCA3, AP3B1, BLOC1S3, BLOC1S6, DKC1, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, OCA2, SFTPB, SFTPC, SLC45A2, TERC, TERT, TINF2, TYR, TYRP1	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	INTERSTITIAL LUNG DISEASE	24	ABCA3, CSF2RA, DKC1, ELMOD2, HPS1, HPS4, ITGA3, NF1, NKX2-1, PARN, RTEL1, SFTPA1, SFTPA2, SFTPB, SFTPC, SLC34A2, SLC7A7, SMPD1, STAT3, TERC, TERT, TINF2, TSC1, TSC2	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	NEONATAL RESPIRATORY DISTRESS - SURFACTANT DYSREGULATION	5	ABCA3, FOXF1, NKX2-1, SFTPB, SFTPC	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	PRIMARY CILIARY DYSKINESIA	32	ARMC4, C21ORF59, CCDC103, CCDC114, CCDC39, CCDC40, CCDC65, CCNO, CENPF, CFTR, DAAAF1, DAAAF2, DAAAF3, DAAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DRC1, DDX1C1, HYDIN, INVS, LRRC6, NME8, OFD1, RPGR, RSPH1, RSPH4A, RSPH9, SPAG1, TSPAN12	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	PULMONARY ARTERY HYPERTENSION (PAH)	11	ACVRL1, BMPR2, CAV1, EIF2AK4, ENG, FOXF1, KCNAS, KCNK3, RASA1, SMAD4, TBX4	Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Whole Exome (WES)	WHOLE EXOME (PANEL 1: PATIENT ONLY)	+/- 23.000	WHOLE EXOME	WES of patient only	1500
Next Generation and Sanger Sequencing Platforms (NGS)	Whole Exome (WES)	WHOLE EXOME (PANEL 2: TRIO OF PATIENT and 2 PARENTS)	+/- 23.000	WHOLE EXOME	WES TRIO analysis of patient and 2 parents to detect de novo variants	2500
Next Generation and Sanger Sequencing Platforms (NGS)	Whole Exome (WES)	WHOLE EXOME (PANEL 3: 2 SIBS and 2 PARENTS)	+/- 23.000	WHOLE EXOME	WES of 2 sibs and 2 parents to detect autosomal recessive variants	2700
Next Generation and Sanger Sequencing Platforms (NGS)	Whole Exome (WES)	WHOLE EXOME (PANEL 4: 3 AFFECTED FAMILY MEMBERS)	+/- 23.000	WHOLE EXOME	WES of 3 affected family members to detect autosomal dominant variants	2700