

## Lista de exames genéticos – Kits de coleta

| DESTAQUES  |                                  |   |
|--|----------------------------------|---|
| Teste  | Tempo de Entrega (dias corridos) | Categoria   |
| Exoma Completo   | 45                               | TODAS   |
| Array (SNP Array de Alta Densidade)  | 45                               | Genética Médica<br>Neurologia                                 |
| Teste da Bochechinha   | 28                               | Genética Médica<br>Neurologia<br>Endocrinologia<br>Nefrologia |
| Painel de Doenças Tratáveis  | 28                               | Genética Médica<br>Neurologia<br>Endocrinologia<br>Nefrologia |
| Sequenciamento Customizado   | 45                               | -----   |
| Pacote Painel de Câncer de Mama e Ovário Hereditários (37 genes) + MLPA BRCA1 + MLPA BRCA2 | 30                               | Oncologia   |

| SEQUENCIAMENTO DE NOVA GERAÇÃO (NGS)<br>PARA DIAGNÓSTICO DE DOENÇAS RARAS           |                                  |                 |
|---|----------------------------------|-----------------|
| Teste   | Tempo de Entrega (dias corridos) | Categoria       |
| Adrenoleucodistrofia (sequenciamento do gene ABCD1)                                 | 45                               | Oncologia       |
| Amiloidose Familiar (sequenciamento do gene TTR)                                    | 30                               | Neurologia      |
| Ataxia Telangiectasia (Sequenciamento do gene ATM)                                  | 45                               | Neurologia      |
| Atrofia Espinhal Progressiva (sequenciamento NGS de SMN1 após MLPA)                 | 45                               | Neurologia      |
| CADASIL (sequenciamento do gene NOTCH3)   | 45                               | Neurologia      |
| Distrofia Muscular de Duchenne (sequenciamento do gene DMD)                         | 30                               | Neurologia      |
| Doença de Wilson (sequenciamento do gene ATP7B)                                     | 45                               | Neurologia      |
| Fibrose Cística (sequenciamento do gene CFTR)                                       | 45                               | Genética Médica |
| Mucopolissacaridose Tipo VII (sequenciamento do gene GUSB)                          | 30                               | Genética Médica |
| Painel Customizado Expandido para Imunodeficiências, Doenças Autoinflamatórias e EI | 45                               | Endocrinologia  |
| Painel de Anemia de Fanconi   | 45                               | Hematologia     |
| Painel de Anemias Hereditárias  | 45                               | Hematologia     |
| Painel de Aneurisma Aórtico   | 45                               | Cardiologia     |
| Painel de Arritmias   | 45                               | Cardiologia     |

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|---|----|--|
| Painel de Ataxias (NGS)   | 45 | Neurologia   |
| Painel de Autismo   | 45 | Genética Médica<br>Neurologia                      |
| Painel de Baixa Estatura  | 45 | Endocrinologia                                     |
| Painel de Câncer Colorretal Hereditário                                   | 30 | Oncologia  |
| Painel de Câncer de Mama e Ovário Hereditários                            | 30 | Oncologia  |
| Painel de Câncer de Próstata  | 45 | Oncologia  |
| Painel de Câncer Hereditário (Completo)                                   | 45 | Oncologia  |
| Painel de Câncer Hereditário (Principais Genes)                           | 30 | Oncologia  |
| Painel de Colestase Crônica   | 45 | Gastroenterologia                                  |
| Painel de Craniosinostoses  | 45 | Genética Médica                                    |
| Painel de Defeitos de Beta-Oxidação de Ácidos Graxos                      | 45 | Neurologia   |
| Painel de Demências e Parkinson   | 45 | Neurologia   |
| Painel de Diabetes Monogênico (MODY)                                      | 45 | Endocrinologia                                     |
| Painel de Discinesia Ciliar Primária                                      | 45 | Cardiologia  |
| Painel de Dislipidemias   | 45 | Cardiologia  |
| Painel de Distonias   | 45 | Neurologia   |
| Painel de Distrofias Musculares, Miopatias e Miastenia                    | 45 | Neurologia   |
| Painel de Distúrbios da Coagulação  | 45 | Hematologia  |
| Painel de Distúrbios da Função Renal                                      | 45 | Nefrologia   |
| Painel de Doença Policística Renal  | 45 | Nefrologia   |
| Painel de Doenças Auto-inflamatórias                                      | 45 | Hematologia  |
| Painel de Doenças Esqueléticas  | 45 | Genética Médica                                    |
| Painel de Doenças Mitocondriais (DNA Nuclear e Mitocondrial)              | 45 | Genética Médica<br>Neurologia<br>Gastroenterologia |
| Painel de Ehlers-Danlos e Cutis Laxa                                      | 45 | Genética Médica                                    |
| Painel de Endocrinopatias Neonatais                                       | 45 | Endocrinologia                                     |
| Painel de Epidermólise Bolhosa  | 45 | Dermatologia                                       |
| Painel de Epilepsias  | 45 | Neurologia   |
| Painel de Esclerose Tuberosa  | 45 | Genética Médica<br>Neurologia                      |
| Painel de Feocromocitoma e Paraganglioma                                  | 45 | Oncologia  |
| Painel de Hemocromatose   | 45 | Hematologia  |
| Painel de Hemofilias A e B  | 45 | Hematologia  |
| Painel de Ictiose e Displasia Ectodérmica                                 | 45 | Dermatologia                                       |
| Painel de Imunodeficiências (Principais Genes)                            | 45 | Hematologia  |

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| SEQUENCIAMENTO DE NOVA GERAÇÃO (NGS)<br>PARA DIAGNÓSTICO DE DOENÇAS RARAS |    |  |
|---|----|--|
| Painel de Imunodeficiências e Doenças Imunológicas (Completo)             | 45 | Hematologia                                    |
| Painel de Infertilidade Masculina   | 45 | Endocrinologia                                 |
| Painel de Leucodistrofias   | 45 | Neurologia                                     |
| Painel de Meningioma  | 45 | Oncologia                                      |
| Painel de Miocardiopatias   | 45 | Cardiologia                                    |
| Painel de Neoplasias Endócrinas   | 45 | Endocrinologia                                 |
| Painel de Neurofibromatose  | 45 | Genética Médica<br>Neurologia                  |
| Painel de Neuropatias   | 45 | Neurologia                                     |
| Painel de Pancreatites  | 45 | Gastroenterologia                              |
| Painel de Paraplegias Espásticas e Esclerose Lateral Amiotrófica          | 45 | Neurologia                                     |
| Painel de Porfirias   | 45 | Hematologia                                    |
| Painel de Retinopatias Hereditárias                                       | 45 | Oftalmologia                                   |
| Painel de Síndrome de Noonan e Rasopatias                                 | 45 | Genética Médica                                |
| Painel de Síndrome Hemolítica-Urêmica                                     | 45 | Nefrologia                                     |
| Painel de Síndrome Nefrótica  | 45 | Nefrologia                                     |
| Painel de Síndromes Clinicamente Reconhecíveis                            | 45 | Genética Médica                                |
| Painel de Stargardt   | 45 | Oftalmologia                                   |
| Painel de Surdez (GJB2/GJB6)  | 45 | Otorrinolaringologia                           |
| Painel de Surdez Hereditária (Expandido)                                  | 45 | Otorrinolaringologia                           |
| Painel de Trombofilias  | 45 | Hematologia                                    |
| Painel Expandido de Melanoma e Câncer de Pele                             | 45 | Oncologia                                      |
| Painel Expandido de Neoplasias Endócrinas                                 | 45 | Endocrinologia                                 |
| Genotipagem de Variante ou Gene (método escolhido pelo sistema)           | 45 | ----   |
| Retinoblastoma (sequenciamento do gene RB1)                               | 30 | Oncologia                                      |
| Sequenciamento de BRCA1, BRCA2 e TP53                                     | 30 | Oncologia                                      |
| Sequenciamento do Gene PTEN   | 30 | Oncologia<br>Neurologia                        |
| Síndrome de Angelman e Prader-Willi (metilação)                           | 75 | Genética Médica                                |
| Síndrome CHARGE (sequenciamento do gene CHD7)                             | 45 | Genética Médica                                |
| Painel para Síndrome de Marfan e Doenças Correlatas                       | 45 | Genética Médica<br>Oftalmologia<br>Cardiologia |
| Síndrome de Marfan (sequenciamento do gene FBN1)                          | 45 | Genética Médica<br>Oftalmologia<br>Cardiologia |
| Síndrome de Rett (sequenciamento do gene MECP2)                           | 45 | Genética Médica<br>Neurologia                  |



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| SEQUENCIAMENTO DE NOVA GERAÇÃO (NGS)<br>PARA DIAGNÓSTICO DE DOENÇAS RARAS |    |                 |
|---|----|-----------------|
| Triagem de Portador de Mutações de Doenças Recessivas                     | 45 | Genética Médica |

| MLPA PARA DOENÇAS RARAS                            |                                  |                 |
|--|----------------------------------|-----------------|
| Teste  | Tempo de Entrega (dias corridos) | Categoria       |
| Atrofia Espinhal Progressiva (MLPA de SMN1 e SMN2) | 30                               | Neurologia      |
| Charcot-Marie-Tooth Tipo 1A e HNPP (MLPA de PMP22) | 75                               | Neurologia      |
| Distrofia Muscular de Duchenne (MLPA do gene DMD)  | 30                               | Neurologia      |
| MLPA de APC  | 45                               | Oncologia       |
| MLPA de ATM  | 45                               | Oncologia       |
| MLPA de BAP1                                       | 45                               | Oncologia       |
| MLPA de BRCA1                                      | 30                               | Oncologia       |
| MLPA de BRCA2                                      | 30                               | Oncologia       |
| MLPA de BRIP1                                      | 45                               | Oncologia       |
| MLPA de CDH1                                       | 45                               | Oncologia       |
| MLPA de CDK4                                       | 45                               | Oncologia       |
| MLPA de CDKN2A                                     | 45                               | Oncologia       |
| MLPA de CHEK2                                      | 45                               | Oncologia       |
| MLPA de EPCAM                                      | 75                               | Oncologia       |
| MLPA de MEN1                                       | 45                               | Oncologia       |
| MLPA de MET  | 45                               | Oncologia       |
| MLPA de MLH1                                       | 45                               | Oncologia       |
| MLPA de MSH2                                       | 45                               | Oncologia       |
| MLPA de MSH6                                       | 45                               | Oncologia       |
| MLPA de MUTYH                                      | 45                               | Oncologia       |
| MLPA de PALB2                                      | 45                               | Oncologia       |
| MLPA de PMS2                                       | 45                               | Oncologia       |
| MLPA de PTEN                                       | 45                               | Oncologia       |
| MLPA de RB1  | 45                               | Oftalmologia    |
| MLPA de RET  | 45                               | Endocrinologia  |
| MLPA de SDHB                                       | 45                               | Oncologia       |
| MLPA de STK11                                      | 45                               | Oncologia       |
| MLPA de TP53                                       | 45                               | Oncologia       |
| MLPA de WT1  | 45                               | Oncologia       |
| Neurofibromatose tipo 1 (MLPA de NF1)              | 45                               | Genética Médica |
| Neurofibromatose tipo 2 (MLPA de NF2)              | 45                               | Genética Médica |

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| MLPA PARA DOENÇAS RARAS                                     |    |                                      |
|---|----|--------------------------------------|
| Síndrome de Alagille (MLPA de JAG1 ou região 20p12)         | 75 | Genética Médica<br>Gastroenterologia |
| Síndrome de Beckwith-Wiedemann (metilação de 11p15)         | 45 | Genética Médica                      |
| Síndrome de Cri du Chat (MLPA da região 5p15)               | 75 | Genética Médica                      |
| Síndrome de Deleção 1p36 (MLPA da região 1p36)              | 75 | Genética Médica                      |
| Síndrome de Langer-Giedion (MLPA da região 8q24)            | 75 | Genética Médica                      |
| Síndrome de Miller-Dieker (MLPA da região 17p13)            | 75 | Genética Médica                      |
| Síndrome de Phelan-McDermid (MLPA da região 22q13)          | 75 | Genética Médica                      |
| Síndrome de Rett (MLPA de MECP2)                            | 75 | Genética Médica<br>Neurologia        |
| Síndrome de Rubinstein-Taybi (MLPA da região 16p13)         | 75 | Genética Médica                      |
| Síndrome de Russell-Silver (metilação de 11p15)             | 45 | Genética Médica                      |
| Síndrome de Smith-Magenis (MLPA da região 17p11)            | 75 | Genética Médica                      |
| Síndrome de Sotos (MLPA da região 5q35)                     | 75 | Genética Médica                      |
| Síndrome de WAGR (MLPA da região 11p13)                     | 75 | Genética Médica                      |
| Síndrome de Williams (MLPA da região 7q11.23)               | 75 | Genética Médica                      |
| Síndrome de Wolf-Hirschhorn (MLPA da região 4p16)           | 75 | Genética Médica                      |
| Síndrome Saethre-Chotzen (MLPA de TWIST1 ou da região 7p21) | 75 | Genética Médica                      |
| Síndrome Velocardiofacial e DiGeorge (MLPA da região 22q11) | 75 | Genética Médica<br>Hematologia       |

| EXPANSÃO DE REPETIÇÕES PARA DOENÇAS RARAS                            |                                  |            |
|--|----------------------------------|------------|
| Teste  | Tempo de Entrega (dias corridos) | Categoria  |
| Ataxia de Friedreich (expansão FXN)                                  | 75                               | Neurologia |
| Ataxia Espinocerebelar Tipo 10 (expansão ATXN10/SCA10)               | 75                               | Neurologia |
| Ataxia Espinocerebelar Tipo 3 - Machado-Joseph (expansão ATXN3/SCA3) | 75                               | Neurologia |
| Ataxia Espinocerebelar Tipo 6 (expansão CACNA1A/SCA6)                | 75                               | Neurologia |
| Ataxia Espinocerebelar Tipo 7 (expansão ATXN7/SCA7)                  | 75                               | Neurologia |
| Ataxias Espinocerebelares por Expansões (SCA1, SCA2, SCA3, SCA6)     | 75                               | Neurologia |
| Distrofia Miotônica Tipo I - Steinert (expansão DMPK)                | 75                               | Neurologia |
| Distrofia Miotônica Tipo II (expansão                                | 75                               | Neurologia |

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| EXPANSÃO DE REPETIÇÕES PARA DOENÇAS RARAS                             |    |                               |
|---|----|-------------------------------|
| CNBP)   |    |                               |
| Doença de Huntington (expansão HTT)                                   | 75 | Neurologia                    |
| Doença de Kennedy (expansão AR)                                       | 75 | Neurologia                    |
| Esclerose Lateral Amiotrófica (expansão C9orf72)                      | 75 | Neurologia                    |
| Hemofilia A (PCR de Inv22 e Inv1)                                     | 75 | Hematologia                   |
| Síndrome de Angelman e Prader-Willi (somente MLPA)                    | 75 | Genética Médica               |
| Síndrome de Hipoventilação Central - Ondine (expansão de gene PHOX2B) | 75 | Neurologia                    |
| Síndrome do X-Frágil (expansão FMR1)                                  | 75 | Genética Médica<br>Neurologia |

| COMPOSIÇÃO DOS PAINÉIS                                      |                  |
|---|------------------|
| Teste   | Genes analisados |
| Adrenoleucodistrofia (sequenciamento do gene ABCD1)         | ABCD1 (1)        |
| Amiloidose Familiar (sequenciamento do gene TTR)            | TTR (1)          |
| Ataxia Telangiectasia (Sequenciamento do gene ATM)          | ATM (1)          |
| CADASIL (sequenciamento do gene NOTCH3)                     | NOTCH3 (1)       |
| Distrofia Muscular de Duchenne (sequenciamento do gene DMD) | DMD (1)          |
| Doença de Wilson (sequenciamento do gene ATP7B)             | ATP7B (1)        |
| Fibrose Cística (sequenciamento do gene CFTR)               | CFTR (1)         |
| Mucopolissacaridose Tipo VII (sequenciamento do gene GUSB)  | GUSB (1)         |



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| COMPOSIÇÃO DOS PAINÉIS  |   |
|---|---|
| Painel Customizado Expandido para Imunodeficiências, Doenças Autoinflamatórias e EI | A2ML1 ABCD4 ACD ACP5 ADA ADA2 ADAM17 ADNP AGA AICDA AIRE AK2 ALG1 ALG12 AP1S3 AP3B1 ARMC4 ARPC1B ATG16L1 ATM B2M BCL10 BCL11B BLM BLNK BRCA1 BRCA2 BRIP1 BTK BUB1B C1QA C1QB C1QC C1R C1S C2 C3 C5 C6 C7 C8A C8B C8orf37 CARD11 CARD14 CARD9 CASP10 CASP8 CAVIN1 CCBE1 CCDC103 CCDC114 CCDC151 CCDC39 CCDC40 CCDC65 CCNO CD19 CD247 CD27 CD3D CD3E CD3G CD40 CD40LG CD55 CD59 CD79A CD79B CD81 CD8A CDCA7 CDSN CDX1 CEBPE CFAP298 CFAP300 CFB CFD CFH CFI CFP CHAMP1 CHD1 CHD7 CIITA CLEC7A CLPB COG6 COG7 CORO1A CR2 CREBBP CRIPT CSF3R CTC1 CTLA4 CTPS1 CXCR4 CYBA CYBB DCLRE1C DEAF1 DEFBI DHFR DKC1 DNAAF1 DNAAF2 DNAAF3 DNAAF4 DNAAF5 DNAH1 DNAH11 DNAH5 DNAI1 DNAI2 DNAJC21 DNAL1 DNASE1L3 DNMT3B DOCK2 DOCK8 DRC1 DSG1 EDA EFL1 EGFR ELANE ELP1 EPG5 ERCC2 ERCC4 ERCC6L2 ETV6 EXTL3 F12 FADD FANCA FANCB FANCC FANCD2 FANCE FANCF FANCG FANCI FANCL FANCM FAS FASLG FAT4 FBXL4 FCGR2C FCGR3A FCN3 FERMT3 FMO3 FOXP1 FOXP3 FUT2 G6PC3 GALNS GAS8 GATA1 GATA2 GATA3 GFI1 GSS GTF2H5 GUCY2C HAX1 HELLS HGSNAT HTR1A HYDIN ICOS IFIH1 IFNGR1 IFNGR2 IGHM IGKC IGLL1 IKBKB IKBKG IKZF1 IKZF2 IL10 IL10RA IL10RB IL12B IL12RB1 IL15 IL15RA IL17A IL17F IL17RA IL17RC IL1RL1 IL1RN IL21 IL21R IL23A IL23R IL2RA IL2RG IL33 IL36RN IL7R IRAK1 IRAK4 IRF2BP2 IRF7 IRF8 IRGM ISG15 ITCH ITGB2 ITK IVD JAGN1 JAK3 KMT2D KRAS LAMTOR2 LAT LCK LEP LIG4 LPIN2 LRBA LRRC6 LRRC8A LYST MAD2L2 MAGT1 MALT1 MAN2B1 MANBA MC2R MCM4 MEFV MGP MMAA MMAB MMACHC MOGS MPL MPO MS4A1 MSN MTHFD1 MUT MVK MYD88 MYO5B NBN NCF1 NCF2 NCF4 NCSTN NFASC NFE2L2 NFKB1 NFKB2 NFKBIA NGF NHEJ1 NHP2 NLRC4 NLRP1 NLRP2 NLRP3 NLRP7 NME8 NOD2 NOP10 NRAS NSMCE3 ORAI1 OXCT1 PALB2 PARN PCCA PCCB PEPD PGM3 PIH1D3 PIK3CD PIK3R1 PLCG2 PMM2 PNP POLA1 POLE PPP1R21 PRDM1 PRF1 PRKCD PRKDC PSENE1 PSMB8 PSTPIP1 PTEN PTPRC RAB27A RAC2 RAD50 RAD51 RAD51C RAG1 RAG2 RASGRP1 RBCK1 RBM8A RELB RFWD3 RFX5 RFXANK RFXAP RNF113A RNF168 RNF186 RNF31 RORC RPL11 RPL15 RPL18 RPL26 RPL35A RPL5 RPS10 RPS17 RPS19 RPS24 RPS26 RPS28 RPS29 RPS7 RPSA RSPH1 RSPH3 RSPH4A RSPH9 RTEL1 SAMD9 SAMD9L SAMHD1 SBDS SCNN1B SCNN1G SDCCAG8 SEMA3E SERAC1 SERPINE2 SERPING1 SGPL1 SH2D1A SH3BP2 SKIV2L SLC29A3 SLC35A1 SLC35A2 SLC35C1 SLC37A4 SLC39A4 SLC39A8 SLC46A1 SLK SLX4 SMARCA1 SMARCD2 SNAI2 SP110 SPAG1 SPATA5 SPINK5 SRP54 SRP72 STAT1 STAT2 STAT3 STAT5B STIM1 STK4 STN1 STX11 STXBP2 TALDO1 TAP1 TAP2 TAPBP TAZ TBCE TBX1 TBXAS1 TCF3 TCN2 TERT TFRC TGFB1 TGFB2 TGFB3 TINF2 TMEM173 TNFAIP3 TNFRSF11A TNFRSF13B TNFRSF13C TNFRSF1A TPI1 TRAC TRAF3IP2 TRNT1 TRPS1 TSR2 TTC25 TTC37 TTC7A TYK2 UBE2T UMPS UNC119 UNC13D UNG USB1 VIPAS39 VPS13B VPS33B VPS45 WAS WIPF1 WRAP53 XIAP XRCC2 ZAP70 ZBTB24 ZMYND10 (436) |
| Painel de Anemia de Fanconi   | BRCA1 BRCA2 BRIP1 ERCC4 FANCA FANCB FANCC FANCD2 FANCE FANCF FANCG FANCI FANCL FANCM PALB2 RAD51 RAD51C SLX4 (18)   |
| Painel de Anemias Hereditárias  | HBB G6PD SLC19A2 SBDS (4)   |

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| COMPOSIÇÃO DOS PAINÉIS                         |   |
|--|---|
| Painel de Aneurisma Aórtico                    | ACTA2 BGN COL1A1 COL1A2 COL3A1 COL5A1 COL5A2 EFEMP2 FBLN5 FBN1 LOX MFAP5 MYH11 MYLK PRKG1 SKI SLC2A10 SMAD3 SMAD6 TGFB2 TGFB3 TGFB1 TGFB2 (23)  |
| Painel de Arritmias                            | ABCC9 ACTN2 AKAP9 ANK2 CACNA1C CACNB2 CALM1 CALM2 CALM3 CASQ2 CAV3 DES DPP6 DSC2 DSG2 DSP EMD GJA5 GPD1L HCN4 JUP KCNA5 KCND3 KCNE1 KCNE2 KCNE3 KCNH2 KCNJ2 KCNJ5 KCNQ1 LMNA MYL4 NKX2-5 NPPA NUP155 PKP2 PLN PRKAG2 RBM20 RYR2 SCN1B SCN2B SCN3B SCN5A SNTA1 TGFB3 TMEM43 TNNI3 TNNT2 TRDN (50)  |
| Painel de Ataxias (NGS)                        | ABHD12 ACO2 AFG3L2 ANO10 APOB APTX ATCAY ATM ATP8A2 BEAN1 CACNA1A CACNA1G CACNB4 CCDC88C CLCN2 CLN5 COQ2 COQ8A CYP27A1 DNMT1 EBF3 ELOVL4 FGF14 FLVCR1 FXN GOSR2 GRM1 ITPR1 KCNA1 KCNC3 KCND3 KCNJ10 KIF1A LAGE3 LAMA1 MRE11 MTTP NPC1 NPC2 NUP107 NUP133 OSGEP PCNA PDSS1 PDSS2 PDYN PEX7 PHYH PMPCA PNKP PNPLA6 POLG PRKCG PTF1A RUBCN SACS SCN2A SETX SIL1 SLC1A3 SLC2A1 SPG7 SPTBN2 SYNE1 SYT14 TDP1 TGM6 TP53RK TPP1 TPRKB TTBK2 TTPA TWNK TXN2 VLDLR WDR4 WDR73 WFS1 WWOX (79) |
| Painel de Autismo                              | ADNP ANKRD11 ARID1B ASH1L AUTS2 CAMK2A CHD2 CHD8 DDX3X DYRK1A EHMT1 FOXP1 GRIA1 GRIN2B HNRNP2 KMT2A KMT2C KMT5B MBD5 MECP2 MED12 NAA15 NEXMIF NLGN3 NLGN4X PACS1 POGZ PPM1D PTCHD1 PTEN RPL10 SCN2A SETD2 SHANK1 SHANK2 SYN1 SYNGAP1 TBL1XR1 TBR1 TRIO TRIP12 UBE3A (42)  |
| Painel de Baixa Estatura                       | ACAN ADAMTS10 ANKRD11 ATR BRAF CBL CCDC8 CDC6 CDT1 CENPJ CEP152 CEP63 CHD7 COL10A1 COL2A1 COL9A1 COL9A2 COMP CREBBP CUL7 FBN1 FGF8 FGFR1 FGFR3 GH1 GHR GHRHR GLI2 GLI3 GNAS HESX1 HRAS IGF1 IGF1R IGF2 IGFALS IHH KRAS LHX3 LHX4 MAP2K1 NPPC NPR2 NRAS OBSL1 ORC1 ORC4 ORC6 OTX2 PAPSS2 PCNT PITX2 POU1F1 PRKAR1A PROP1 PTH1R PTPN11 RAF1 RBBP8 SHOC2 SHOX SOS1 SOX3 SOX9 SRCAP STAT5B XRCC4 (67)   |
| Painel de Câncer Colorretal Hereditário        | APC ATM BARD1 BLM BRCA1 BRCA2 BRIP1 CDH1 CDK4 CDKN2A CHEK2 EGFR EPCAM FANCC MEN1 MET MLH1 MSH2 MSH6 MUTYH NBN NF1 NF2 PALB2 PIK3CA PMS2 POLD1 POLE PTEN RAD51C RAD51D RB1 RECQL RET STK11 TP53 WT1 (37)   |
| Painel de Câncer de Mama e Ovário Hereditários | APC ATM BARD1 BLM BRCA1 BRCA2 BRIP1 CDH1 CDK4 CDKN2A CHEK2 EGFR EPCAM FANCC MEN1 MET MLH1 MSH2 MSH6 MUTYH NBN NF1 NF2 PALB2 PIK3CA PMS2 POLD1 POLE PTEN RAD51C RAD51D RB1 RECQL RET STK11 TP53 WT1 (37)   |
| Painel de Câncer de Próstata                   | ATM BRCA1 BRCA2 CDK12 CHEK1 CHEK2 FANCA MLH1 MSH2 MSH6 PALB2 PMS2 RAD51C (13)   |



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| COMPOSIÇÃO DOS PAINÉIS                               |   |
|--|---|
| Painel de Câncer Hereditário (Completo)              | ACD AIP AKT1 ALK ANKRD26 APC ARMC5 ASCL1 ASXL1 ATM ATP4A ATR AXIN2 BAP1 BARD1 BDNF BLM BMPR1A BPGM BRAF BRCA1 BRCA2 BRIP1 BUB1B CABLES1 CASP10 CASP9 CBL CD70 CDC73 CDH1 CDH23 CDK4 CDKN1B CDKN1C CDKN2A CEBPA CEP57 CHEK2 CREBBP CSF3R CTC1 CTNNA1 CTNNB1 CTR9 CYLD DDB2 DDX41 DICER1 DIS3L2 DKC1 DLST DNAJC21 DNMT3B DOCK8 EDN3 EFL1 EGFR EGLN1 EGLN2 EPAS1 EPCAM ERCC2 ERCC3 ERCC4 ERCC5 ERCC6 ERCC6L2 ETV6 EXT1 EXT2 EZH2 FAN1 FANCA FANCB FANCC FANCD2 FANCE FANCF FANGC FANCI FANCL FANCM FAS FASLG FBXW7 FGFR1 FH FIBP FLCN FOXE1 G6PC GATA1 GATA2 GLMN GNAS GPC3 HCLS1 HIF3A HNF1A HNF1B HOXB13 HRAS IPMK JAG1 JAK2 KDM1A KDM3B KIF1B KIT KLLN KRAS LAPTM5 LDAH LIG4 LZTR1 MAD2L2 MAGT1 MAP2K1 MAP2K2 MAP3K1 MAX MCM4 MDH2 MEN1 MET MTF MLH1 MLH3 MMP1 MNX1 MRE11 MSH2 MSH3 MSH6 MSR1 MTAP MUTYH MYCN NBN NF1 NF2 NHP2 NME1 NOP10 NRAS NSD1 NTHL1 NTRK1 NYN1RIN OS9 PALB2 PARN PAX5 PBRM1 PDGFB PDGFRA PDGFRB PHOX2B PIK3CA PMS2 POLD1 POLE POLH POT1 PPP2R3B PRF1 PRKAR1A PSMC3IP PTCH1 PTCH2 PTEN PTPN11 RAD50 RAD51 RAD51C RAD51D RAD54L RAF1 RASA2 RASAL1 RB1 RBBP6 RECQL RECQL4 RET RFW3 RHBDF2 RMI2 RNASL1 RNF139 RRAS RSP1 RTEL1 RUNX1 SAMD9 SAMD9L SASH1 SBDS SDHA SDHAF2 SDHB SDHC SDHD SEC23B SERPINA1 SETBP1 SH2B3 SH2D1A SHOC2 SLC25A11 SLX4 SMAD4 SMARCA4 SMARCA1 SMARCB1 SMARCE1 SOS1 SPRN SRP54 SRP72 STAT3 STK11 SUFU TERC TERF2IP TERT TET2 TEX15 TGFB2 THSD1 TINF2 TMC6 TMC8 TMEM127 TOP3A TP53 TPCN2 TRIM28 TRIP13 TSC1 TSC2 UBE2T USP8 VHL WAS WIPF1 WNT10A WRAP53 WRN WT1 WWOX XIAP XPA XPC XRCC2 ZNF687 (257) |
| Painel de Câncer Hereditário (Principais Genes)      | APC ATM BARD1 BLM BRCA1 BRCA2 BRIP1 CDH1 CDK4 CDKN2A CHEK2 EGFR EPCAM FANCC MEN1 MET MLH1 MSH2 MSH6 MUTYH NBN NF1 NF2 PALB2 PIK3CA PMS2 POLD1 POLE PTEN RAD51C RAD51D RB1 RECQL RET STK11 TP53 WT1 (37)   |
| Painel de Colestase Crônica                          | ABCB11 ABCB4 ATP8B1 CFTR KIF12 LSR MYO5B NR1H4 PPM1F SERPINA1 TJP2 USP53 VIPAS39 VPS33B WDR83OS (15)  |
| Painel de Craniosinostoses                           | FGFR1 FGFR2 FGFR3 TWIST1 (4)  |
| Painel de Defeitos de Beta-Oxidação de Ácidos Graxos | ACADM ACADVL CPT1A CPT2 HADHA HADHB SLC25A20 (7)  |
| Painel de Demências e Parkinson                      | ABCD1 APP ARSA ATP13A2 ATP1A3 ATP7B CHMP2B CSF1R CYP27A1 DCTN1 DNAJC6 EIF4G1 FBXO7 FUS GALC GBA GCH1 GFAP GLA GRN HEXA HTRA2 ITM2B LMNB1 LRRK2 MAPT NOTCH3 NPC1 NPC2 PANK2 PARK7 PINK1 PLA2G6 PNKD POLG PPT1 PRKN PRKRA PRNP PRRT2 PSAP PSEN1 PSEN2 SGCE SLC2A1 SLC6A3 SNCA SPG11 SPR SQSTM1 TARDBP TH THAP1 TOR1A TREM2 TYROBP UBQLN2 UCHL1 VCP VPS35 (60)   |
| Painel de Diabetes Monogênico (MODY)                 | ABCC8 APPL1 BLK CEL EIF2AK3 GATA6 GCK HNF1A HNF1B HNF4A INS KCNJ11 KLF11 NEUROD1 NEUROG3 PAX4 PDX1 PLAGL1 PTF1A RFX6 SH2B1 SLC19A2 SLC2A2 WFS1 ZFP57 (25)   |

## Lista de exames genéticos – Kits de coleta

| COMPOSIÇÃO DOS PAINÉIS                                 |  |
|--|--|
| Painel de Discinesia Ciliar Primária                   | ARMC4 CCDC103 CCDC114 CCDC151 CCDC39 CCDC40 CCDC65 CCNO CFAP298 CFAP300 DNAAF1 DNAAF2 DNAAF3 DNAAF4 DNAAF5 DNAH1 DNAH11 DNAH5 DNAI1 DNAI2 DNAJB13 DNAL1 DRC1 GAS8 HYDIN LRRC6 NME8 PIH1D3 RSPH1 RSPH3 RSPH4A RSPH9 SPAG1 TTC25 ZMYND10 (35)  |
| Painel de Dislipidemias                                | LDLR PCSK9 APOB LDLRAP1 (4)  |
| Painel de Distonias                                    | ADCY5 ANO3 ARG1 ARSA ATM ATP1A3 ATP7B CACNA1B COL6A3 CP GCDH GCH1 GNAL HPRT1 KCNMA1 KCTD17 MRE11 PANK2 PCNA PLA2G6 PNKD PRKN PRKRA PRRT2 RELN SGCE SLC2A1 SLC6A3 SPR TAF1 TH THAP1 TIMM8A TOR1A TUBB4A WDR45 (36)  |
| Painel de Distrofias Musculares, Miopatias e Miastenia | ACTA1 AGRN ANO5 B3GALNT2 B4GAT1 BAG3 BIN1 CAPN3 CAV3 CFL2 CHAT CHKB CHRNA1 CHRNB1 CHRND CHRNE CNTN1 COL6A1 COL6A2 COL6A3 COLQ CPT2 CRYAB DAG1 DES DMD DNAJB6 DNM2 DOK7 DPAGT1 DPM1 DPM3 DYSF EMD FHL1 FKRP FKTN FLNC GAA GFPT1 GMPPB GNE HNRNPA1 HNRNPA2B1 IGHMBP2 ITGA7 KBTBD13 LAMA2 LARGE1 LDB3 LMNA MAGEL2 MATR3 MTM1 MUSK MYH7 MYL2 MYOT NEB ORAI1 PABPN1 PLEC PNPLA2 POMGNT1 POMGNT2 POMK POMT1 POMT2 PYGM RAPSN RXYL1 RYR1 SELENON SGCA SGCB SGCD SGCG SQSTM1 STIM1 TCAP TGFB1 TIA1 TK2 TNNT1 TPM2 TPM3 TRIM32 TTN VCP VMA21 YARS2 (91) |
| Painel de Distúrbios da Coagulação                     | ACTN1 ANO6 AP3B1 ARPC1B BLOC1S3 COL4A1 COL4A2 DTNBP1 F10 F11 F12 F13A1 F13B F2 F5 F7 F8 F9 FGA FGB FGG FLI1 GF11B GGCX GP1BA GP1BB GP6 GP9 HPS1 HPS3 HPS4 HPS5 HPS6 ITGA2 ITGA2B ITGB3 LMAN1 MCFD2 NBEAL2 P2RY12 PLA2G4A PLAT PLAU PRKACG RASGRP2 SERPIND1 SERPINE1 SLFN14 TBXA2R TBXAS1 VKORC1 VWF (52)   |
| Painel de Distúrbios da Função Renal                   | AQP2 ATP6V0A4 ATP6V1B1 AVPR2 BSND CLCNKA CLCNKB CLDN16 CLDN19 CNNM2 CTNS GLA KCNJ1 SCNN1A SCNN1B SCNN1G SLC12A1 SLC12A3 SLC4A4 TRPM6 (20)  |
| Painel de Doença Policística Renal                     | DNAJB11 DZIP1L GANAB NOTCH2 PKD1 PKD2 PKHD1 (7)  |
| Painel de Doenças Auto-inflamatórias                   | ADAM17 ARPC1B CARD11 CD55 EGFR FOXP3 IL10RA IL10RB IL1RN IL36RN ITCH LPIN2 MEFV MVK NLRC4 NLRP12 NLRP3 NOD2 PLCG2 PSTPIP1 TNFAIP3 TNFRSF1A (22)  |



## Lista de exames genéticos – Kits de coleta

| COMPOSIÇÃO DOS PAINÉIS                                       |   |
|--|---|
| Painel de Doenças Esqueléticas                               | <p>ABL1 ACAN ACP5 ACVR1 ADAMTSL2 AGA AGPS ALPL ALX1 ALX3 ALX4 AMER1 ANKH ANO5 ARCN1 ARSB ARSE ASCC1 ATP6V0A2 B3GALT6 B3GAT3 B4GALT7 BGN BHLHA9 BMP1 BMP2 BMPER BMPR1B C12orf57 CA2 CANT1 CASR CCDC8 CCN6 CDC45 CDC6 CDKN1C CDT1 CEP120 CHST11 CHST3 CHSY1 CLCN7 COG1 COL10A1 COL11A1 COL11A2 COL1A1 COL1A2 COL2A1 COL9A1 COL9A2 COL9A3 COMP CREB3L1 CRTAP CTSA CTSK CUL7 CYP26B1 CYP27B1 CYP2R1 CICN5 DDR2 DDX58 DHCR24 DHODH DIP2C DLL3 DLX3 DLX5 DMP1 DVL1 DVL3 DYM DYNC2H1 DYNC2LI1 EBP EDNRA EFN1 EFTUD2 EHHADH EIF2AK3 EIF4A3 ENPP1 ESCO2 EVC EVC2 EXT1 EXT2 FAM111A FAM20C FAM98C FBLN1 FBN1 FERMT3 FGF16 FGF23 FGF9 FGFR1 FGFR2 FGFR3 FIG4 FKBP10 FLNA FLNB FN1 FNDC3B FUCA1 FZD2 GALNS GDF3 GDF5 GDF6 GJA1 GLB1 GLI1 GLI3 GMNN GNAS GNPAT GNPTAB GNPTG GNS GORAB GPC6 GPX4 GUSB HDAC4 HDAC6 HES7 HGSNAT HNF4A HNRNPA1 HNRNPA2B1 HOXA11 HOXA13 HOXD13 HPGD HSPG2 ICK IDS IDUA IFIH1 IFITM5 IFT122 IFT140 IFT172 IFT43 IFT52 IFT80 IFT81 IHH IMPAD1 INPPL1 INTU IQCE KAT6B KIAA0586 KIF22 LBR LEMD3 LFNG LIFR LMNA LMX1B LONP1 LRP4 LRP5 LTBP3 MAFB MAN2B1 MAP3K20 MAP3K7 MATN3 MBTPS1 MBTPS2 MCM3 MCM5 MCM7 MECOM MEOX1 MESP2 MMP13 MMP2 MMP9 MNX1 MSX2 MYH3 MYO18B MYT1 NAGLU NANS NEK1 NEU1 NKX3-2 NOG NOTCH2 NPR2 NSDHL NT5E NUDT6 OBSL1 ORC1 ORC4 ORC6 OSTM1 P3H1 P4HB PAM16 PAPSS2 PCGF2 PCNT PCYT1A PDE4D PEX5 PEX7 PHEX PIK3C2A PLEKHM1 PLOD2 PLS3 POLR1A POP1 POR PPIB PPP3CA PRG4 PRKAR1A PTDSS1 PTH1R PTHLH PTPN11 PYCR1 RAB33B RBBP8 RECQL4 RIN1 RIPPLY2 RMRP RNU4ATAC ROR2 RSPO2 RSPRY1 RUNX2 SBDS SEC23A SEC24D SERPINF1 SERPINH1 SF3B4 SFRP4 SGSH SH3PXD2B SHOX SIK3 SLC10A7 SLC17A5 SLC26A2 SLC29A3 SLC34A1 SLC34A3 SLC35D1 SLC39A13 SLC05A1 SMAD4 SMARCAL1 SNRPB SNX10 SOST SOX9 SP7 SPARC SQSTM1 SUCO SULF1 SUMF1 TBCE TBX15 TBX3 TBX4 TBX6 TBXAS1 TCIRG1 TCTEX1D2 TENT5A TGFB1 TMCO1 TMEM256 TMEM38B TNFRSF11A TNFRSF11B TNFSF11 TONSL TRAPPC2 TREM2 TRIP11 TRIP4 TRPS1 TRPV4 TTC21B TYROBP UNC45A VCP VDR WDR19 WDR34 WDR35 WDR60 WNT1 WNT10B WNT3 WNT5A WNT7A XYLT1 XYLT2 ZBTB16 ZMPSTE24 ZNF141 ZNF687 ZSWIM6 (321)</p> |
| Painel de Doenças Mitocondriais (DNA Nuclear e Mitocondrial) | <p>AARS2 ACAD9 AIFM1 APOPT1 ATP5F1A ATP5F1E ATPAF2 BCS1L BOLA3 C12orf65 MICOS13 C1QBP CARS2 CHCHD10 COX10 COX14 COX15 COX20 COX6B1 CYC1 DARS2 DGUOK DNA2 EARS2 ECHS1 ELAC2 FARS2 FASTKD2 FBXL4 FDX2 FDXR FOXRED1 GFER GFM1 GTPBP3 HADHA HADHB IBA57 ISCA1 ISCA2 ISCU LIAS LIPT2 LRPPRC LYRM4 LYRM7 MARS2 MGME1 MPC1 MPV17 MRPL3 MRPL44 MRPS16 MRPS2 MRPS22 MRPS34 MRPS7 MSTO1 MT-ATP6 MT-ATP8 MT-CO1 MT-CO2 MT-CO3 MT-CYB MT-ND1 MT-ND2 MT-ND3 MT-ND4 MT-ND4L MT-ND5 MT-ND6 MT-RNR1 MT-TA MT-TC MT-TD MT-TE MT-TF MT-TG MT-TH MT-TI MT-TK MT-TL1 MT-TL2 MT-TM MT-TN MT-TP MT-TQ MT-TR MT-TS1 MT-TS2 MT-TV MT-TW MT-TY MTFMT MTO1 NARS2 NDUFA1 NDUFA10 NDUFA11 NDUFA12 NDUFA2 NDUFA9 NDUFAF1 NDUFAF2 NDUFAF3 NDUFAF4 NDUFAF5 NDUFAF6 NDUFB3 NDUFB8 NDUFB9 NDUFS1 NDUFS2 NDUFS3 NDUFS4 NDUFS6 NDUFS7 NDUFS8 NDUFV1 NDUFV2</p>   |



## Lista de exames genéticos – Kits de coleta

| COMPOSIÇÃO DOS PAINÉIS               |   |
|--------------------------------------|---|
|                                      | NFU1 NUBPL OPA1 PCK2 PET100 PNPLA8 PNPT1 POLG POLG2 PUS1 RMND1 RNASEH1 RRM2B SCO1 SDHA SDHAF1 SDHD SFXN4 SLC25A26 SLC25A3 SLC25A4 SUCLA2 SUCLG1 SUOX SURF1 TACO1 TANGO2 TARS2 TIMMDC1 TK2 TMEM126B TMEM70 TRIT1 TRMT10C TRMT5 TSFM TTC19 TUFM TWNK TXN2 TYMP UQCC2 UQCC3 UQCRB UQCRC2 UQCRQ VARS2 WARS2 YARS2 (169)   |
| Painel de Doenças Tratáveis          | <p>           ABCB11 ABCB4 ABCC8 ABCD1 ABCD4 ACADM ACADVL ACAT1 ADA ADAMTS13 AGL AICDA AK2 AKR1D1 ALDH7A1 ALDOB ALPL AMACR APOA5 APOC2 AQP2 ARG1 ARSA ARSB ASL ASS1 ATP6V0A4 ATP6V1B1 ATP7A ATP7B ATP8B1 AVPR2 BAAT BCKDHA BCKDHB BCKDK BCL10 BLNK BSND BTB BTK CAD CARD11 CASR CBS CD247 CD320 CD3D CD3E CD3G CD40 CD40LG CD79A CD79B CDCA8 CFTR CIITA CLCNKA CLCNKB CLDN16 CLDN19 CNNM2 CORO1A CPS1 CPT1A CPT2 CTNS CTPS1 CXCR2 CXCR4 CYBA CYBB CYP11B1 CYP11B2 CYP17A1 CYP27A1 CYP27B1 CYP2R1 CYP7A1 CYP7B1 DBT DCLRE1C DDC DGAT1 DLD DMD DNAJC12 DNAJC21 DOCK2 DUOX2 DUOX2A2 EFL1 ELANE ETFA ETFB ETFDH ETHE1 F8 F9 FAH FBPI FGA FLAD1 FOLR1 FOXA2 FOXE1 FOXN1 FOXP3 G6PC G6PC3 GAA GALE GALK1 GALNS GALT GAMT GATA2 GATM GBA GBE1 GCDH GCH1 GCK GF1 GGCX GJB2 GJB6 GLA GLB1 GLIS3 GLUD1 GOT2 GPIHBP1 GUSB GYS2 HADH HADHA HADHB HAX1 HBB HESX1 HLCS HMGCL HMGCS2 HPD HSD3B2 HSD3B7 HYOU1 IDS IDUA IFNGR1 IFNGR2 IGLL1 IGSF1 IKBKB IL12B IL12RB1 IL2RA IL2RG IL7R INS INSR IRF8 IRS4 IVD IYD JAGN1 JAK3 KCNJ1 KCNJ11 LAT LCK LCT LHX3 LHX4 LIPA LMBRD1 LMF1 LPL LRRC8A MAGT1 MALT1 MAP3K14 MC2R MMAA MMAB MMACHC MMADHC MOCS1 MPI MPL MPO MRAP MTHFR MTR MTRR MTTP MUT MYD88 MYH9 NAGLU NAGS NCF2 NCF4 NEUROG3 NHEJ1 NKX2-1 NKX2-5 NNT NPC1 NPC2 NR0B1 ORAI1 OTC OTX2 OXCT1 PAH PAX8 PCBD1 PCCA PCCB PDXK PGM1 PHEX PHGDH PHKA2 PHKB PIK3R1 PLPBP PNP PNPO POU1F1 PRF1 PRKDC PROP1 PSAT1 PSPH PTPRC PTS PYGL QDPR RAC2 RAG1 RAG2 RASGRP1 RB1 RFX5 RFXANK RFXAP RORC SAMD9 SBDS SCNN1A SCNN1B SCNN1G SGSH SH2D1A SI SLC12A1 SLC16A1 SLC19A2 SLC19A3 SLC22A5 SLC25A13 SLC25A15 SLC25A19 SLC25A20 SLC26A3 SLC26A4 SLC26A7 SLC27A5 SLC2A1 SLC2A2 SLC37A4 SLC39A4 SLC46A1 SLC52A2 SLC52A3 SLC5A1 SLC5A5 SLC6A6 SLC7A7 SLC7A9 SMN1 SMPD1 SOX3 SPR SRP54 STAR STAT1 STX11 STXBP2 TANGO2 TAP1 TAP2 TAPBP TAT TBL1X TCN2 TFRC TG TH THRA TJP2 TPK1 TPO TPP1 TRH TRHR TRPM6 TSHB TSHR TTPA TUBB1 UGT1A1 UNC13D UNG USP53 VDR VKORC1 VPS45 WAS WIPF1 XIAP ZAP70 (325)         </p> |
| Painel de Ehlers-Danlos e Cutis Laxa | <p>           ADAMTS2 AEBP1 ALDH18A1 ATP6V0A2 ATP6V1A ATP6V1E1 ATP7A B3GALT6 B3GAT3 B4GALT7 CHST14 COL1A1 COL1A2 COL3A1 COL5A1 COL5A2 EFEMP2 ELN FBLN5 FKBP14 FLNA GORAB GZF1 HRAS KIF22 LTBP4 PIK3R1 PLOD1 PPP1CB PYCR1 RIN2 SLC2A10 SLC39A13 TNXB (34)         </p>   |
| Painel de Endocrinopatias Neonatais  | <p>           ABCC8 CYP11B1 CYP17A1 DUOX2 GCK GLIS3 GLUD1 HADH INSR IYD KCNJ11 LHX4 NR0B1 PAX8 POU1F1 PROP1 SLC16A1 SLC2A2 SLC5A5 TG THRA THRB TPO TSHB (25)         </p>   |
| Painel de Epidermólise Bolhosa       | <p>           CAST CD151 COL17A1 COL7A1 DSP DST FERMT1 ITGA3 ITGA6 ITGB4 KRT14 KRT5 LAMA3 LAMB3 LAMC2 PLEC TGM5 (17)         </p>   |



## Lista de exames genéticos – Kits de coleta

| COMPOSIÇÃO DOS PAINÉIS                         |  |
|--|--|
| Painel de Epilepsias                           | AARS1 ACER3 ADAM22 ADGRV1 ADRA2B ADSL AIMP2 ALDH7A1 ALG13 AMT AP3B2 ARHGEF9 ARV1 ARX ASAH1 ATP13A2 ATP1A2 ATP6V1A ATP7A ATP8A2 BRAF BRAT1 BSCL2 CACNA1A CACNA1D CACNA1E CACNB4 CASK CASR CCDC88A CDK5 CDKL5 CERS1 CHD2 CHRNA2 CHRNA4 CHRNB2 CLCN2 CLDN5 CLN3 CLN5 CLN6 CLN8 CLTC CNNM2 CNPY3 CNTN2 CNTNAP2 CPA6 CPLX1 CSTB CTSD CYFIP2 DCX DEAF1 DENND5A DEPDC5 DHDDS DIAPH1 DIP2A DLAT DNAJC5 DNM1 DOCK7 EEF1A2 EIF2S3 EMX2 EPM2A EXT2 FOLR1 FOXG1 FRRS1L GABBR2 GABRA1 GABRA2 GABRA3 GABRA5 GABRB1 GABRB2 GABRB3 GABRG2 GAMT GATM GBA GCSH GLDC GNAO1 GOSR2 GPAA1 GRIA4 GRIN1 GRIN2A GRIN2B GRIN2D GRN HACE1 HCN1 HECW2 HEXA HEXB HNRNPU ICK IER3IP1 IQSEC2 ITPA KANSL1 KATNB1 KCNA1 KCNA2 KCNB1 KCNC1 KCNJ10 KCNMA1 KCNQ2 KCNQ3 KCNT1 KCNT2 KCTD17 KCTD3 KCTD7 LAMB1 LGI1 LIAS LMNB2 MBD5 MDH2 MECP2 MED17 MEF2C MFSD8 MOCS1 MOCS2 NACC1 NDE1 NECAP1 NHLRC1 NPC1 NPC2 NPRL2 NPRL3 NR4A2 NRXN1 NTRK2 NUS1 OTUD6B PACS2 PAFAH1B1 PCDH12 PCDH19 PDHA1 PDHX PDP1 PIGA PIGC PIGN PIGP PIGT PLAA PLCB1 PLPBP PNKP PNPO POLG POLG2 PPP3CA PPT1 PRDM8 PRICKLE1 PRICKLE2 PRRT2 PTPN23 QARS1 RAB11A RBFOX1 RELN RHOBTB2 ROGDI ROXB RPH3A RTN4IP1 RTTN SARS1 SCARB2 SCN1A SCN1B SCN2A SCN3A SCN8A SCN9A SGCE SHH SIK1 SIX3 SLC12A5 SLC13A5 SLC25A22 SLC2A1 SLC35A3 SLC45A1 SLC6A1 SLC6A8 SLC6A9 SLC9A6 SMC1A SMS SNAP25 SNIP1 SPATA5 SPTAN1 SRPX2 ST3GAL3 ST3GAL5 STRADA STX1B STXBP1 SUOX SYN1 SYNGAP1 SYNJ1 SZT2 TBC1D24 TBCD TCF4 TMTC3 TPP1 TRIO TSC1 TSC2 TUBA1A UBA5 UBE3A UFC1 UFM1 VARS1 VRK2 WASF1 WDR45B WWOX YWHAG ZEB2 (240) |
| Painel de Esclerose Tuberosa                   | TSC1 TSC2 (2)  |
| Painel de Feocromocitoma e Paraganglioma       | VHL SDHA SDHB SDHC SDHD SDHAF2 MAX TMEM127 RET NF1 (10)  |
| Painel de Hemocromatose                        | HFE HFE2 HAMP SLC40A1 TFR2 (5)   |
| Painel de Hemofilias A e B                     | F8 F9 (2)  |
| Painel de Ictiose e Displasia Ectodérmica      | ABCA12 ABHD5 ALDH3A2 ALOX12B ALOXE3 AP1S1 CDH1 CDH3 CLDN1 COG6 CSTA CYP4F22 DLX3 EDA EDAR EDARADD ELOVL4 ERCC2 FLG GJA1 GJB2 GJB6 GRHL2 HOXC13 HR IFT122 IKBKG ITPR2 JUP KDF1 KREMEN1 KRT1 KRT10 KRT14 KRT2 KRT74 KRT85 LIPN LOR MBTPS2 MSX1 NECTIN1 NECTIN4 NFKBIA NIPAL4 NLRP1 PKP1 PNPLA1 POMP PRKD1 SLC27A4 SMARCAD1 SNAP29 ST14 STS TGM1 TP63 TWIST2 WNT10A (59)  |
| Painel de Imunodeficiências (Principais Genes) | ADA AICDA BLNK BTK CD247 CD3D CD3E CD3G CD40 CD40LG CD79A CD79B CIITA CYBA CYBB DCLRE1C ELANE FOXN1 FOXP3 G6PC3 GATA2 GF11 HAX1 IFNGR1 IFNGR2 IGLL1 IL12RB1 IL2RG IL7R JAK3 LRRC8A MAGT1 MPO MYD88 NCF1 NCF2 NCF4 NHEJ1 ORAI1 PNP PRF1 PTPRC RAC2 RAG1 RAG2 RFX5 RFXANK RFXAP SH2D1A STAT1 STX11 STXBP2 TAP1 TAP2 TAPBP UNC13D UNG WAS WIPF1 XIAP (60)   |



## Lista de exames genéticos – Kits de coleta

| COMPOSIÇÃO DOS PAINÉIS  |  |
|---|--|
| Painel de Imunodeficiências e Doenças Imunológicas (Completo) | A2ML1 ABCD4 ACD ACP5 ADA ADA2 ADAM17 ADNP AGA AICDA AIRE AK2 ALG1 ALG12 AP3B1 ARMC4 ARPC1B ATM B2M BCL10 BCL11B BLM BLNK BRCA1 BRCA2 BRIP1 BTK BUB1B C1QA C1QB C1QC C1R C1S C2 C3 C5 C6 C7 C8A C8B C8orf37 CARD11 CARD9 CASP10 CASP8 CAVIN1 CCBE1 CCDC103 CCDC114 CCDC151 CCDC39 CCDC40 CCDC65 CCNO CD19 CD247 CD27 CD3D CD3E CD3G CD40 CD40LG CD55 CD59 CD79A CD79B CD81 CD8A CDCA7 CDSN CEBPE CFAP298 CFAP300 CFB CFD CFH CFI CFP CHAMP1 CHD1 CHD7 CIITA CLEC7A CLPB COG6 COG7 CORO1A CPN1 CR2 CREBBP CRIPT CSF3R CTC1 CTLA4 CTPS1 CXCR4 CYBA CYBB DCLRE1C DEAF1 DHFR DKC1 DNAAF1 DNAAF2 DNAAF3 DNAAF4 DNAAF5 DNAH1 DNAH11 DNAH5 DNAI1 DNAI2 DNAJC21 DNAL1 DNASE1L3 DNMT3B DOCK2 DOCK8 DRC1 DSG1 EFL1 EGFR ELANE ELP1 EPG5 ERCC2 ERCC4 ERCC6L2 ETV6 EXTL3 F12 FADD FANCA FANCB FANCC FANCD2 FANCE FANCF FANCG FANCI FANCL FANCM FAS FASLG FAT4 FBXL4 FCGR3A FCN3 FERMT3 FMO3 FOXP1 FOXP3 G6PC3 GALNS GAS8 GATA1 GATA2 GF11 GSS GTF2H5 HAX1 HELLS HGSNAT HTR1A HYDIN ICOS IFIH1 IFNGR1 IFNGR2 IGHM IGKC IGLL1 IKBKB IKBKG IKZF1 IL10RA IL10RB IL12B IL12RB1 IL17F IL17RA IL17RC IL1RN IL21 IL21R IL2RA IL2RG IL36RN IL7R IRAK4 IRF2BP2 IRF7 IRF8 ISG15 ITCH ITGB2 ITK IVD JAGN1 JAK3 KMT2D KRAS LAMTOR2 LAT LCK LEP LIG4 LPIN2 LRBA LRRC6 LRRC8A LYST MAD2L2 MAGT1 MALT1 MAN2B1 MANBA MC2R MCM4 MEFV MGP MMAA MMAB MMACHC MOGS MPL MPO MS4A1 MSN MTHFD1 MUT MVK MYD88 NBN NCF1 NCF2 NCF4 NFASC NFE2L2 NFKB1 NFKB2 NFKBIA NGF NHEJ1 NHP2 NLR4 NLRP12 NLRP3 NME8 NOD2 NOP10 NRAS NSMCE3 ORAI1 OXCT1 PALB2 PARN PCCA PCCB PEPD PGM3 PIH1D3 PIK3CD PIK3R1 PLCG2 PMM2 PNP POLA1 POLE PPP1R21 PRF1 PRKCD PRKDC PSTPIP1 PTPRC RAB27A RAC2 RAD50 RAD51 RAD51C RAG1 RAG2 RASGRP1 RBCK1 RBM8A RELB RFX5 RFXANK RFXAP RNF113A RNF168 RNF31 RORC RPL11 RPL15 RPL18 RPL26 RPL35A RPL5 RPS10 RPS17 RPS19 RPS24 RPS26 RPS28 RPS29 RPS7 RPSA RSPH1 RSPH3 RSPH4A RSPH9 RTEL1 SAMD9 SAMD9L SAMHD1 SBDS SCNN1B SCNN1G SDCCAG8 SEMA3E SERAC1 SERPING1 SGPL1 SH2D1A SKIV2L SLC35A1 SLC35A2 SLC35C1 SLC37A4 SLC39A4 SLC39A8 SLC46A1 SLK SLX4 SMARCAL1 SMARCD2 SNAI2 SP110 SPAG1 SPATA5 SPINK5 SRP72 STAT1 STAT2 STAT3 STAT5B STIM1 STK4 STN1 STX11 STXBP2 TALDO1 TAP1 TAP2 TAPBP TAZ TBCE TBX1 TBXAS1 TCN2 TERT TFRC TGFB3 TINF2 TMEM173 TNFAIP3 TNFRSF13B TNFRSF13C TNFRSF1A TPI1 TRAC TRAF3IP2 TRNT1 TRPS1 TSR2 TTC25 TTC37 TTC7A TYK2 UBE2T UMPS UNC119 UNC13D UNG USB1 VIPAS39 VPS13B VPS33B VPS45 WAS WIPF1 WRAP53 XIAP XRCC2 ZAP70 ZBTB24 ZMYND10 (399) |
| Painel de Infertilidade Masculina                             | AK7 ARMC2 AURKC CATSPER1 CATSPER2 CDC14A CEP19 CFAP43 CFAP44 CFAP69 CFTR DNAH1 DNAH6 DPY19L2 FANCM FSIP2 KLHL10 MEIOB NANOS1 NR5A1 PMFBP1 QRIH2 SLC26A8 SOHLH1 SOX8 SPATA16 SPINK2 SUN5 SYCP3 TDRD9 TEX11 TEX14 TEX15 TSGA10 USP9Y WDR66 XRCC2 (37)  |
| Painel de Leucodistrofias                                     | AARS2 ABCD1 ACOX1 ADAR AIMP1 ALDH3A2 ARSA ASPA ATP7A ATP7B ATPAF2 BCAP31 BCS1L CLCN2 COL4A1 COQ2 COQ8A COQ9 COX10  |



## Lista de exames genéticos – Kits de coleta

| COMPOSIÇÃO DOS PAINÉIS          |   |
|---------------------------------|---|
|                                 | COX15 CSF1R CYP27A1 CYP2U1 CYP7B1 D2HGDH DARS1 DARS2 DGUOK EARS2 EIF2B1 EIF2B2 EIF2B3 EIF2B4 EIF2B5 ERCC2 ERCC3 ERCC6 ERCC8 ETFDH FA2H FAM126A FUCA1 GALC GBE1 GFAP GFM1 GJA1 GJC2 GLA GLB1 GM2A GTF2H5 HEPACAM HEXA HEXB HSD17B4 HSPD1 HTRA1 L2HGDH LAMA2 LMNB1 MCOLN1 MLC1 MPLKIP MRPS16 MTFMT NDUFAF1 NDUFS1 NDUFS2 NDUFS4 NDUFS7 NDUFS8 NDUFV1 NOTCH3 NPC1 NPC2 OCLN OCRL PEX1 PEX10 PEX11B PEX12 PEX13 PEX14 PEX16 PEX19 PEX2 PEX26 PEX3 PEX5 PEX6 PEX7 PHGDH PHYH PLP1 POLG POLG2 POLR3A POLR3B PPT1 PRF1 PSAP PSAT1 RNASEH2A RNASEH2B RNASEH2C RNASET2 RRM2B SAMHD1 SCO1 SCP2 SDHA SDHAF1 SDHB SLC16A2 SLC17A5 SLC25A1 SLC25A12 SLC25A4 SOX10 SPART SPAST SPG11 SPG21 SPG7 STX11 STXBP2 SUCLA2 SUMF1 SURF1 TACO1 TREX1 TUBB4A TUFM TWNK TYMP TYROBP UNC13D ZFYVE26 (139)   |
| Painel de Meningioma            | ARMC5 BAP1 LZTR1 PDGFB PTEN SMARCB1 SMARCE1 SUFU (8)  |
| Painel de Miocardiopatias       | AARS2 ABCC6 ABCC9 ACAD8 ACAD9 ACADVL ACTA1 ACTC1 ACTN2 ADCY5 AGK AGL AHCY ALG1 ALG12 ALMS1 ALPK3 ANKS6 APOPT1 ARSB ATAD3A ATP5F1E ATPAF2 BAG3 BCS1L BMP2 BOLA3 BRAF BSCL2 C1QBP CACNA1C CALR3 CAV3 CAVIN1 CENPE CEP19 CHKB CLIC2 CLN3 COA5 COA6 COQ2 COQ4 COX10 COX14 COX15 COX20 COX6B1 COX7B CPT1A CPT2 CRYAB CSRP3 D2HGDH DCAF8 DES DLD DMD DNAJC19 DOLK DPM3 DPP6 DSG2 DSP DTNA ECHS1 ELAC2 EMD EPG5 ERBB3 EYA4 FAH FASTKD2 FBXL4 FHL1 FIG4 FKRP FKTN FLAD1 FLNC FOXRED1 FTO FUCA1 FXN GAA GATAD1 GBE1 GLA GLB1 GMPPB GNPTAB GNS GPC3 GSN GTPBP3 GYS1 HADH HADHA HADHB HCCS HGSNAT HPS1 HRAS HSD17B10 IDH2 IDUA ITPA JPH2 JUP KCNE1 KCNH1 KCNJ2 KCNQ1 LAMA4 LAMP2 LDB3 LIAS LMNA MAP2K1 MAP2K2 MCCC2 MGME1 MIB1 MLYCD MRPL3 MRPL4 MRPS22 MRPS7 MTFMT MTO1 MUT MYBPC3 MYH6 MYH7 MYL2 MYL3 MYLK2 MYO6 MYOT MYOZ2 MYPN NAGLU NDUFA1 NDUFA10 NDUFA11 NDUFA12 NDUFA2 NDUFA6 NDUFA9 NDUFAF1 NDUFAF2 NDUFAF3 NDUFAF4 NDUFAF5 NDUFAF6 NDUFB11 NDUFB3 NDUFB8 NDUFB9 NDUFS1 NDUFS2 NDUFS3 NDUFS4 NDUFS6 NDUFS7 NDUFS8 NDUFV1 NDUFV2 NEU1 NEXN NKX2-5 NONO NUBPL PAM16 PCCA PCCB PET100 PGM1 PHYH PIGT PLN PMM2 PNPLA2 POLG POMT1 PPCS PRDM16 PRG4 PRKAG2 PRKARIA PSEN1 PSEN2 PSMB4 PSMB8 PSMB9 RAB3GAP2 RAF1 RBCK1 RBM20 RIT1 RMND1 RYR2 SCN5A SCO1 SCO2 SDHA SDHAF1 SDHD SELENON SGCA SGCB SGCD SGCG SGSH SHOC2 SLC19A2 SLC22A5 SLC25A20 SLC25A26 SLC25A3 SLC25A4 SOS1 SPEG SURF1 SYNE1 SYNE2 TACO1 TANGO2 TAPT1 TAZ TBX1 TBX3 TBX5 TCAP TF TIMMDC1 TMEM126A TMEM126B TMEM43 TMEM70 TMPO TNNC1 TNNI3 TNNI3K TNNT2 TOP3A TPM1 TPM3 TRIT1 TRMT5 TRNT1 TSC1 TSFM TTN TTR TWNK UBR1 VCL VPS33A WFS1 XK XPNPEP3 YARS2 (263) |
| Painel de Neoplasias Endócrinas | APC ATM BARD1 BLM BRCA1 BRCA2 BRIP1 CDH1 CDK4 CDKN2A CHEK2 EGFR EPCAM FANCC MEN1 MET MLH1 MSH2 MSH6 MUTYH NBN NF1 NF2 PALB2 PIK3CA PMS2 POLD1 POLE PTEN RAD51C RAD51D RB1 RECQL RET STK11 TP53 WT1 (37)   |
| Painel de Neurofibromatose      | NF1 NF2 SPRED1 (3)  |



## Lista de exames genéticos – Kits de coleta

| COMPOSIÇÃO DOS PAINÉIS   |   |
|--|---|
| Painel de Neuropatias  | AAAS AARS1 ABHD12 AIFM1 AP1S1 ATL1 ATL3 ATP1A1 ATP7A BSCL2 C12orf65 CCT5 COA7 COX6A1 CTD1P1 DCAF8 DHH DHTKD1 DMXL2 DNAJB2 DNM2 DNMT1 DST DYNC1H1 EGR2 ELP1 EXOC4 FBLN5 FGD4 FIG4 GAN GARS1 GBE1 GDAP1 GJB1 GNB4 GSN HARS1 HINT1 HK1 HSPB1 HSPB8 IARS2 IGHMBP2 INF2 JPH1 KARS1 KIF1A KIF1B KLC2 LITAF LMNA LRSAM1 MARS1 MCM3AP MED25 MFN2 MORC2 MPZ MTMR2 MYH14 NAGLU NDRG1 NEFH NEFL NGF OPA1 PDK3 PLEKHG5 PMP22 POLG PRDM12 PRPS1 PRX RAB7A RETREG1 RNF170 SBF1 SBF2 SCN10A SCN11A SCN9A SCO2 SCP2 SH3TC2 SIGMAR1 SLC12A6 SLC25A46 SNAP29 SOX10 SPG11 SPTBN4 SPTLC1 SPTLC2 SURF1 TBCE TDP1 TFG TRIM2 TRPV4 TTR VCP WNK1 YARS1 (104)  |
| Painel de Pancreatites   | APOA5 APOC2 CFTR CTCR GPIHBP1 LMF1 LPL PRSS1 SPINK1 (9)   |
| Painel de Paraplegias Espásticas e Esclerose Lateral Amiotrófica | ABCD1 ALS2 ANG AP4B1 AP4E1 AP4M1 AP4S1 AP5Z1 ARG1 ATL1 BSCL2 C12ORF65 CHMP2B CYP7B1 DYNC1H1 ERBB4 ERLIN2 FA2H FIG4 FUS GARS1 GJC2 HSPB1 HSPB8 HSPD1 IGHMBP2 KIF1A KIF5A L1CAM MATR3 NEK1 NIPA1 OPTN PARK7 PFN1 PLP1 PNPLA6 REEP1 RNF170 RTN2 SACS SETX SIGMAR1 SLC33A1 SOD1 SPART SPAST SPG11 SPG21 SPG7 SQSTM1 TARDBP TBK1 TP73 TRPV4 UBQLN2 VAPB VCP WASHC5 ZFYVE26 (60)  |
| Painel de Porfirias  | ALAD ALAS2 CPOX FECH HMBS PPOX UROD UROS (8)  |
| Painel de Retinopatias Hereditárias                              | ABCA4 ABCC6 ABCD1 ABHD12 ACO2 ADAM9 ADGRV1 AHI1 AHR AIPL1 ALMS1 AMACR ARL13B ARL6 ATF6 B9D1 B9D2 BBIP1 BBS1 BBS10 BBS12 BBS2 BBS4 BBS5 BBS7 BBS9 BEST1 C12ORF65 C1QTNF5 C8ORF37 CA4 CABP4 CACNA1F CACNA2D4 CASK CC2D2A CDH23 CDH3 CDHR1 CEP250 CEP290 CEP41 CEP78 CERKL CFH CHM CIB2 CISD2 CLN3 CLN5 CLN6 CLN8 CLRN1 CNGA1 CNGA3 CNGB1 CNGB3 CNNM4 CPLANE1 CRB1 CRX CTNNA1 CTSD CYP4V2 WHRN DHDDS DHX38 DNAJC5 EFEMP1 ELOVL4 EYS FAM161A FLVCR1 FRMD7 FSCN2 FZD4 GDF6 GJB2 GJB6 GNAT1 GNAT2 GNPTG GPR143 GPR179 GRK1 GRM6 GRN GUCA1A GUCA1B GUCY2D HGSNAT HK1 HMCN1 HMX1 IDH3B IFT140 IFT27 IMPDH1 IMPG1 IMPG2 INPP5E IQCB1 ITM2B KCNJ13 KCNV2 KCTD7 KIF7 KLHL7 LAMA1 LCA5 LRAT LRIT3 LRP5 LZTFL1 MAK MERTK MFN2 MFRP MFSD8 MKKS MKS1 MMACHC MVK MYO7A NDP NEUROD1 NMNAT1 NPHP1 NPHP3 NPHP4 NR2F1 NRL NYX OAT OFD1 OPA1 OPA3 OPN1LW OTX2 PAX6 PCARE PCDH15 PDE6A PDE6B PDE6C PDE6G PDE6H PDZD7 PEX1 PEX10 PEX11B PEX12 PEX13 PEX14 PEX16 PEX19 PEX2 PEX26 PEX3 PEX5 PEX6 PEX7 PHYH PITPNM3 PNPLA6 POC5 PPT1 PRCD PROM1 PRPF3 PRPF31 PRPF6 PRPF8 PRPH2 PRPS1 RAB28 RAX2 RBP3 RBP4 RD3 RDH12 RDH5 RGR RGS9 RGS9BP RHO RIMS1 RLBP1 ROM1 RP1 RP1L1 RP2 RP9 RPE65 RPGR RPGRIP1 RPGRIP1L RS1 SAG SDCCAG8 SEMA4A SLC24A1 SNRNP200 SPATA7 TCTN1 TCTN2 TEAD1 TIMM8A TIMP3 TMEM126A TMEM138 TMEM216 TMEM237 TMEM67 TOPORS TPP1 TRAPPC3 TRIM32 TRPM1 TSPAN12 TTC21B TTC8 TUBGCP4 TUBGCP6 TULP1 TYR UNC119 USH1C USH1G USH2A VPS13B WDPCP WDR19 WFS1 WHRN ZNF408 ZNF513 HARS1 (238) |
| Painel de Síndrome de Noonan e Rasopatias                        | A2ML1 BRAF CBL HRAS KRAS LZTR1 MAP2K1 MAP2K2 NF1 NRAS PPP1CB PTPN11 RAF1 RASA1 RASA2 RIT1 RRAS SHOC2 SOS1 SOS2 SPRED1 (21)  |



## Lista de exames genéticos – Kits de coleta

| COMPOSIÇÃO DOS PAINÉIS                         |  |
|--|--|
| Painel de Síndrome Hemolítica-Urêmica          | C3 CD46 CFB CFH CFI THBD (6)   |
| Painel de Síndrome Nefrótica                   | ACTN4 ADGRE1 APOL1 ARHGAP24 ARHGDIS ARHGEF17 AVIL CD2AP COL4A3 COL4A4 COL4A5 COQ2 COQ6 COQ8B DGKE EMP2 FN1 IFIH1 IL36G INF2 ITGA3 KANK1 KANK2 LAMB2 LMX1B MAGI2 MYH9 MYO1E NPHS1 NPHS2 NUP107 NUP133 NUP160 NUP205 NUP85 NUP93 PAX2 PDSS2 PLCE1 PMM2 PTPRO SCARB2 SGPL1 SLC17A5 SMARCAL1 TBC1D8B TRPC6 WNK4 WT1 XPO5 (50)  |
| Painel de Síndromes Clinicamente Reconhecíveis | A2ML1 ABCC9 ACTB ACTG1 ALMS1 ANKRD11 AP3B1 AP3D1 ARHGAP31 ARID1A ARID1B ARID2 ARL6 ARL6IP1 ASXL1 ATR ATRX BANF1 BBIP1 BBS1 BBS10 BBS12 BBS2 BBS4 BBS5 BBS7 BBS9 BLM BLOC1S3 BLOC1S6 BRAF CBL CCDC65 CCDC8 CCDC88A CCDC88C CD96 CDC6 CDT1 CENPJ CEP152 CEP290 CEP63 CHD7 CHRNG CNTNAP2 CREBBP CUL7 DHCR7 DHODH DLL4 DOCK6 DTNBP1 DVL1 DVL3 EDN3 EDNRB EHMT1 ELP4 EOGT EP300 EPG5 ERCC6 ERCC6L2 ERCC8 EYA1 EZH2 FBN1 FBN2 FGD1 FOXL2 GATA3 GCM2 GLE1 GNAS GPC3 GPR143 GRHL3 HDAC8 HNF1A HNF1B HPS1 HPS3 HPS4 HPS5 HPS6 HRAS HSPG2 IFT27 IRF6 JAG1 KAT6B KDM6A KIF1BP KIF7 KIT KITLG KMT2A KMT2C KMT2D KRAS KRIT1 LMNA LMX1B LRMDA LYST LZTFL1 LZTR1 MAP2K1 MAP2K2 MATR3 MED12 MID1 MITF MKKS MKS1 MLPH MYBPC1 MYH3 MYH8 MYO5A NF1 NFIX NIN NIPBL NOTCH1 NOTCH2 NRAS NRXN1 NSD1 NSMCE2 OBSL1 OCA2 OFD1 ORC1 ORC4 ORC6 PAX3 PAX6 PHF6 PIEZO2 POLR1C POLR1D PPP1CB PSMC3IP PTCH1 PTCH2 PTEN PTH PTH1R PTHLH PTPN11 RAB27A RAD21 RAF1 RAI1 RASA1 RASA2 RBBP8 RBM8A RBPJ RECQL4 RIT1 RNF113A RNF125 RNF135 RNF139 RNF168 RNF170 ROR2 RPS6KA3 RRAS SALL1 SALL4 SDCCAG8 SERPINF1 SETBP1 SF3B4 SHOC2 SIX5 SKI SKIV2L SLC24A5 SLC25A24 SLC45A2 SMAD3 SMAD4 SMARCA2 SMARCA4 SMARCB1 SMARCE1 SMC1A SMC3 SNAI2 SOS1 SOS2 SOX10 SPECC1L SPRED1 SRCAP STX16 SUFU TBCE TBX5 TCF4 TCOF1 TFAP2A TGFB2 TGFB3 TGFB1 TGFB2 TMEM67 TNNT2 TNNT3 TP63 TPM2 TRAP1 TRIM32 TRIT1 TRPS1 TTC8 TYR TYROBP TYR1 UBR1 VIPAS39 VKORC1 VPS13B VPS33B WASHC5 WDPCP WNT5A WRN ZEB2 ZMPSTE24 ZNF141 ZNF148 (237) |
| Painel de Stargardt                            | ABCA4 ELOVL4 PROM1 PRPH2 (4)   |
| Painel de Surdez (GJB2/GJB6)                   | GJB2 GJB6 (2)  |
| Painel de Surdez Hereditária (Expandido)       | ACTB ACTG1 ADGRV1 ATP6V0A4 ATP6V1B1 BCS1L BSND CATSPER2 CCDC50 CDH23 CEACAM16 CEMIP CLDN14 CLRN1 COCH COL11A2 COL9A2 COL9A3 CRYM WHRN DIAPH1 DSPP ECE1 EDNRA EDNRB ERCC2 ERCC3 ESPN ESRRB EYA4 FAS FGF3 FGFR3 GATA3 GIPC3 GJA1 GJB1 GJB2 GJB3 GJB4 GJB6 GPSM2 GRHL2 GRXCR1 GSDME HGF ILDR1 JAG1 KCNE1 KCNJ10 KCNQ1 KCNQ4 LHFPL5 LHX3 LOXHD1 LRTOMT MARVELD2 MITF MSRB3 MTAP MYH14 MYH9 MYO15A MYO3A MYO6 MYO7A MYOC NDP NR2F1 OTOA OTOF PAX3 PCDH15 PDZD7 PJKV PMP22 POU3F4 POU4F3 PRPS1 PTPRO RDX SERPINB6 SIX1 SIX5 SLC17A8 SLC26A4 SLC26A5 SLC4A11 SMPX SNAI2 SOX2 SPINK5 STRC TBL1X TECTA TIMM8A TJP2 TMC1 TMIE TMPRSS3 TPRN TRIOBP USH1C USH1G USH2A WFS1 WHRN (107)  |



## Lista de exames genéticos – Kits de coleta

| COMPOSIÇÃO DOS PAINÉIS                              |  |
|---|--|
| Painel de Trombofilias                              | PROC PROS1 F5 F2 SERPINC1 ADAMTS13 CBS (7)   |
| Painel Expandido de Melanoma e Câncer de Pele       | ACD ATM BAP1 BARD1 BLM BRCA1 BRCA2 BRIP1 CDK4 CDKN2A CHEK2 CYLD DDB2 EPCAM ERCC2 ERCC3 ERCC4 ERCC5 ERCC6 FH FLCN GLMN MLH1 MSH2 MSH6 PALB2 PMS2 POLD1 POLE POLH POT1 PTCH1 RAD51C RAD51D RECQL RSP01 TERF2IP TGFBR1 TMC6 TMC8 TP53 XPA XPC (43)  |
| Painel Expandido de Neoplasias Endócrinas           | AIP AKT1 APC ARMC5 ATM BARD1 BLM BRCA1 BRCA2 BRIP1 CDH1 CDH23 CDK4 CDKN1B CDKN2A CHEK2 DICER1 EGFR EPCAM FANCC GPR101 KIF1B MAX MEN1 MET MLH1 MSH2 MSH6 MUTYH NBN NF1 NF2 PALB2 PIK3CA PMS2 POLD1 POLE PRKAR1A PTEN RAD51C RAD51D RB1 RECQL RET SDHA SDHAF2 SDHB SDHC SDHD SEC23B SMARCA4 STK11 TMEM127 TP53 VHL WT1 (56)  |
| Painel para Síndrome de Marfan e Doenças Correlatas | ACTA2 ADAMTS2 ADAMTSL4 AEBP1 ALDH18A1 ATP6V0A2 ATP6V1A ATP6V1E1 ATP7A B3GALT6 B3GAT3 B4GALT7 BGN CBS CHST14 COL11A1 COL11A2 COL1A1 COL1A2 COL2A1 COL3A1 COL5A1 COL5A2 COL9A1 COL9A2 EFEMP2 ELN FBLN5 FBN1 FBN2 FKBP14 FLNA FOXE3 GORAB GZF1 HRAS KIF22 LOX LTBP2 LTBP3 LTBP4 MFAP5 MYH11 MYLK PIK3R1 PLOD1 PPP1CB PRKG1 PYCR1 RIN2 ROBO4 SKI SLC2A10 SLC39A13 SMAD3 SMAD6 TGFB2 TGFB3 TGFBR1 TGFBR2 TNXB (61)  |
| Retinoblastoma (sequenciamento do gene RB1)         | RB1 (1)  |
| Sequenciamento de BRCA1, BRCA2 e TP53               | BRCA1 BRCA2 TP53 (3)   |
| Sequenciamento do Gene PTEN                         | PTEN (1)   |
| Síndrome CHARGE (sequenciamento do gene CHD7)       | CHD7 (1)   |
| Síndrome de Marfan (sequenciamento do gene FBN1)    | FBN1 (1)   |
| Teste da Bochechinha                                | ABCB11 ABCB4 ABCC8 ABCD1 ABCD4 ACADM ACADVL ACAT1 ADA ADAMTS13 AGL AICDA AK2 AKR1D1 ALDH7A1 ALDOB ALPL AMACR APOA5 APOC2 AQP2 ARG1 ARSA ARSB ASL ASS1 ATP6V0A4 ATP6V1B1 ATP7A ATP7B ATP8B1 AVPR2 BAAT BCKDHA BCKDHB BCKDK BCL10 BLNK BSND BTB BTK CAD CARD11 CASR CBS CD247 CD320 CD3D CD3E CD3G CD40 CD40LG CD79A CD79B CDCA8 CFTR CIITA CLCNKA CLCNKB CLDN16 CLDN19 CNM2 CORO1A CPS1 CPT1A CPT2 CTNS CTPS1 CXCR2 CXCR4 CYBA CYBB CYP11B1 CYP11B2 CYP17A1 CYP27A1 CYP27B1 CYP2R1 CYP7A1 CYP7B1 DBT DCLRE1C DDC DGAT1 DLD DMD DNAJC12 DNAJC21 DOCK2 DUOX2 DUOX2A EFL1 ELANE ETFA ETFB ETFDH ETHE1 F8 F9 FAH FBPI FGA FLAD1 FOLR1 FOXA2 FOXE1 FOXN1 FOXP3 G6PC G6PC3 GALE GALK1 GALNS GALT GAMT GATA2 GATM GBA GBE1 GCDH GCH1 GCK GF1 GGCX GJB2 GJB6 GLIS3 GLUD1 GOT2 GPIHBP1 GUSB GYS2 HADH HADHA HADHB HAX1 HBB HESX1 HLCS HMGCL HMGCS2 HPD HSD3B2 HSD3B7 HYOU1 IDS IDUA IFNGR1 IFNGR2 IGLL1 IGSF1 IKBKB IL12B IL12RB1 IL2RA IL2RG IL7R INS INSR IRF8 IRS4 IVD IYD JAGN1 JAK3 KCNJ1 KCNJ11 LAT LCK LCT LHX3 LHX4 LIPA LMBRD1 LMF1 LPL LRRC8A MAGT1 MALT1 MAP3K14 MC2R MMAA MMAB |

## Lista de exames genéticos – Kits de coleta

| COMPOSIÇÃO DOS PAINÉIS                                |  |
|---|--|
|   | MMACHC MMADHC MOCS1 MPI MPL MPO MRAP MTHFR MTR MTRR MTTT MUT MYD88 MYH9 NAGS NCF2 NCF4 NEUROG3 NHEJ1 NKX2-1 NKX2-5 NNT NR0B1 ORAI1 OTC OTX2 OXCT1 PAH PAX8 PCBD1 PCCA PCCB PDXK PGM1 PHEX PHGDH PHKA2 PHKB PIK3R1 PLPBP PNP PNPO POU1F1 PRF1 PRKDC PROP1 PSAT1 PSPH PTPRC PTS PYGL QDPR RAC2 RAG1 RAG2 RASGRP1 RB1 RFX5 RFXANK RFXAP RORC SAMD9 SBDS SCNN1A SCNN1B SCNN1G SH2D1A SI SLC12A1 SLC16A1 SLC19A2 SLC19A3 SLC22A5 SLC25A13 SLC25A15 SLC25A19 SLC25A20 SLC26A3 SLC26A4 SLC26A7 SLC27A5 SLC2A1 SLC2A2 SLC37A4 SLC39A4 SLC46A1 SLC52A2 SLC52A3 SLC5A1 SLC5A5 SLC6A6 SLC7A7 SLC7A9 SMN1 SOX3 SPR SRP54 STAR STAT1 STX11 STXBP2 TANGO2 TAP1 TAP2 TAPBP TAT TBL1X TCN2 TFRC TG TH THRA TJP2 TPK1 TPO TPP1 TRH TRHR TRPM6 TSHB TSHR TTPA TUBB1 UGT1A1 UNC13D UNG USP53 VDR VKORC1 VPS45 WAS WIPF1 XIAP ZAP70 (317)  |
| Triagem de Portador de Mutações de Doenças Recessivas | ABCC8 ACADM ACADVL ADA ADAMTS2 AGA AGL AGXT AIRE ALDH3A2 ALDOB ALG6 ALMS1 ALPL AMT ARG1 ARSA ASL ASPA ASS1 ATM ATP7B BBS1 BBS10 BBS12 BBS2 BCKDHA BCKDHB BCS1L BLM BTD CAPN3 CBS CFTR CLN3 CLN5 CLN6 CLN8 CLRN1 COL4A3 COL4A4 CPS1 CPT1A CPT2 CTNS CTSK CYP11B1 CYP27A1 DBT DHCR7 DHDDS DLD DYSF ELP1 ERCC6 ERCC8 EVC EVC2 F11 FAH FANCA FANCC FKRP FKTN G6PC GAA GALT GALK1 GALT GBA GCDH GJB2 GLB1 GLDC GNE GNPTAB GNPTG GRHPR HADHA HBB HEXA HEXB HGSNAT HLCS HMGCL HOGA1 HSD17B4 HYLS1 IDUA IVD KCNJ11 LAMA2 LAMA3 LAMB3 LAMC2 LIPA LRPPRC MAN2B1 MCOLN1 MESP2 MKS1 MLC1 MMAA MMAB MMACHC MPI MPL MTTT MUT MYO7A NAGLU NBN NEB NPC1 NPC2 NPHS1 NPHS2 OPA3 PAH PC PCCA PCCB PCDH15 PEX1 PEX10 PEX12 PEX2 PEX6 PEX7 PHGDH PKHD1 PMM2 POMGNT1 PPT1 PROP1 PTS RTEL1 SACS SGCA SGCB SGCD SGCG SGSH SLC12A6 SLC17A5 SLC22A5 SLC26A2 SLC26A4 SLC35A3 SLC37A4 SMPD1 STAR SUMF1 TAT TCIRG1 TGM1 TH TMEM216 TPP1 TTPA USH1C USH2A VPS13B XPA XPC ZFYVE26 (166) |

O Laboratório de Análise Genômica se reserva o direito de alterar a composição de qualquer dado painel sem aviso prévio, seguindo critérios técnico-científicos.