DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL DG-4.3.0 (21 GENES)

| Gene | Twist X2 covered 10x | Twist X2 covered 20x | srWGS covered 10x | srWGS covered 15x | srWGS covered 20x | Associated Phenotype description and OMIM disease ID |
|---------|----------------------|----------------------|-------------------|-------------------|-------------------|---|
| ACD | 100% | 100% | 100% | 99.9% | 98.9% | ?Dyskeratosis congenita, autosomal recessive 7, 616553;?Dyskeratosis congenita, autosomal dominant 6, 616553 |
| CTC1 | 100% | 100% | 100% | 99.9% | 98.6% | Cerebroretinal microangiopathy with calcifications and cysts, 612199 |
| DCLRE1B | 100% | 100% | 100% | 99.9% | 98.3% | Dyskeratosis congenita, autosomal recessive 8, 620133 |
| DKC1 | 100% | 100% | 99.1% | 89.3% | 71% | ?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000 |

| GRHL2 | 100% | 100% | 100% | 99.9% | 99.2% | Deafness, autosomal dominant 28, 608641;Ectodermal dysplasia/short stature syndrome, 616029;Corneal dystrophy, posterior polymorphous, 4, 618031 |
|-------|-------|-------|------|-------|-------|---|
| LIG4 | 100% | 100% | 100% | 100% | 99.7% | LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500 |
| NAF1 | 100% | 100% | 100% | 100% | 98.8% | Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 7, 620365 |
| NHP2 | 100% | 100% | 100% | 99.8% | 97.8% | Dyskeratosis congenita, autosomal recessive 2, 613987 |
| NOP10 | 92.5% | 92.5% | 100% | 100% | 98.8% | ?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9, 620400;?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2, 620425;?Dyskeratosis congenita, autosomal recessive 1, 224230 |

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|------|-------|-------|------|-------|-------|---|
| NPM1 | 87.6% | 87.6% | 100% | 99.8% | 99.2% | Leukemia, acute myeloid, somatic, 601626 |
| PARN | 97.5% | 95.8% | 100% | 100% | 99.5% | Dyskeratosis congenita, autosomal recessive 6, 616353;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere- related, 4, 616371 |
| POT1 | 100% | 100% | 100% | 100% | 99.7% | Tumor predisposition syndrome 3, 615848;?Cerebroretinal microangiopathy with calcifications and cysts 3, 620368;?Pulmonary fibrosis and/or bone marrow failure syndrome, telomererelated, 8, 620367 |
| RPA1 | 100% | 100% | 100% | 99.9% | 98.9% | Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 6, 619767 |

| RTEL1 | 100% | 100% | 100% | 99.9% | 98.7% | Dyskeratosis congenita, autosomal dominant 4, 615190;Dyskeratosis congenita, autosomal recessive 5, 615190;Pulmonary fibrosis and/or bone marrow failure syndrome, telomererelated, 3, 616373 |
|---------|-------|------|------|-------|-------|--|
| TERC | | | | | | Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 2, 614743;Dyskeratosis congenita, autosomal dominant 1, 127550 |
| TERF2IP | 99.9% | 95% | 100% | 100% | 98.4% | |
| TERT | 100% | 100% | 100% | 99.8% | 97.8% | Dyskeratosis congenita, autosomal dominant 2, 613989;Dyskeratosis congenita, autosomal recessive 4, 613989;Pulmonary fibrosis and/or bone marrow failure syndrome, telomererelated, 1, 614742;{Melanoma, cutaneous malignant, 9}, 615134;{Leukemia, acute myeloid}, 601626 |

| TINF2 | 100% | 100% | 100% | 100% | 98.9% | Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130 |
|--------|-------|-------|------|-------|-------|--|
| USB1 | 94.9% | 93.2% | 100% | 100% | 98.9% | Poikiloderma with neutropenia, 604173 |
| WRAP53 | 100% | 100% | 100% | 99.9% | 97.7% | Dyskeratosis congenita, autosomal recessive 3, 613988 |
| ZCCHC8 | 100% | 100% | 100% | 99.9% | 99.3% | ?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 5, 618674 |

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.

TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.

srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38. srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38. srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38. non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions: November 25th, 2024.

This list is accurate for panel version DG 4.2.0

Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors