

WES DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA

DG 3.5

| Gene | Median coverage | % covered >10x | % covered >20x | OMIM disease ID |
|--------|-----------------|----------------|----------------|----------------------|
| ACD | 120.5 | 100.0 | 100.0 | 616553 |
| CTC1 | 120.1 | 100.0 | 100.0 | 612199 |
| DKC1 | 105.8 | 100.0 | 100.0 | 305000 |
| GRHL2 | 131.5 | 100.0 | 100.0 | 616029 |
| LIG4 | 156.5 | 100.0 | 100.0 | 606593 |
| NHP2 | 118.6 | 100.0 | 100.0 | 613987 |
| NOP10 | 131.5 | 100.0 | 100.0 | 224230 |
| NPM1 | 143.1 | 100.0 | 100.0 | - |
| PARN | 132.6 | 97.0 | 95.9 | 616353 |
| POT1 | 156.3 | 100.0 | 100.0 | 615848 |
| RPA1 | 129.5 | 100.0 | 100.0 | 619767 |
| RTEL1 | 126.6 | 100.0 | 100.0 | 615190 |
| TERC | | | | 127550;614743 |
| TERT | 156.5 | 100.0 | 100.0 | 615134;614742;613989 |
| TINF2 | 124.5 | 100.0 | 100.0 | 268130;613990 |
| USB1 | 132.2 | 100.0 | 100.0 | 604173 |
| WRAP53 | 132.0 | 100.0 | 100.0 | 613988 |

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 is the default chemistry for all WES samples. Agilent V5 was the default chemistry until Q3 2021.

Median Coverage describes the average number of reads seen across 50 exomes.

Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.

Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.

Genes with no value for coverage are non protein coding genes.

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: September 1st, 2021.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors