

# WES DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA

## DG 2.16

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
ACD	159.6	100.0	100.0	616553
CTC1	105.5	100.0	99.3	612199
DKC1	91.2	99.8	97.7	305000
GRHL2	116.8	100.0	100.0	616029
LIG4	173.4	100.0	99.8	606593
NHP2	121.9	100.0	99.2	613987
NOP10	120.5	100.0	100.0	224230
PARN	127.3	99.9	99.5	616353
POT1	97.7	99.9	98.5	615848
RTEL1	131.1	99.7	97.7	615190
TERC				614743;127550
TERT	144.1	99.7	97.6	613989;615134;614742
TINF2	177.1	100.0	100.0	613990;268130
USB1	118.2	99.8	97.2	604173
WRAP53	162.8	100.0	100.0	613988

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

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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : October 1st, 2016.

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