

# WES DYSKERATOSIS CONGENITA DG 2.14

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
ACD	135.2	100.0	98.2	616553
CTC1	119.0	100.0	99.8	612199
DKC1	111.9	99.6	98.1	305000
GRHL2	134.6	100.0	100.0	616029
LIG4	165.6	100.0	99.6	606593
NHP2	111.0	100.0	100.0	613987
NOP10	160.5	100.0	100.0	224230
PARN	128.4	99.9	98.0	616353
POT1	90.7	99.6	96.0	615848
RTEL1	110.9	99.2	95.1	615190
TERC				614743;127550
TERT	138.3	95.3	92.0	613989;615134;614742
TINF2	184.0	100.0	100.0	613990;268130
USB1	125.0	99.9	98.2	604173
WRAP53	154.4	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.

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