WES DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA

DG 3.1

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
ACD	148.6	100.0	99.9	616553
CTC1	128.0	100.0	99.6	612199
DKC1	108.6	99.8	98.7	305000
GRHL2	139.4	100.0	100.0	616029
LIG4	222.9	100.0	99.9	606593
NHP2	146.1	100.0	100.0	613987
NOP10	147.4	100.0	99.8	224230
NPM1	90.1	98.2	85.3	-
PARN	134.2	81.2	81.1	616353
POT1	120.7	99.9	99.0	615848
RTEL1	127.7	99.5	96.8	615190
TERC				127550;614743
TERT	132.4	96.2	94.5	615134;614742;613989
TINF2	187.5	100.0	100.0	268130;613990
USB1	139.6	100.0	99.4	604173
WRAP53	187.5	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 10x 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