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OCULAR MALFORMATIONS GENE PANEL - TECHNICAL INFORMATION

Design: The Ocular Malformations gene panel was designed as part of a custom probe set from Twist Bioscience (ID TE-98169478) to cover 41 genes associated with congenital eye malformations (see list below). The panel includes all coding exons as reported on RefSeq, Ensembl, CCDS, Gencode and CytoBand, and their immediate intronic sequences (+/- 15bp). The final design encompassed 100% of the target regions.

Method: Library preparation and target enrichment was performed using the TE-98169478 probe set (Twist Bioscience) and Nextera Flex for Enrichment (Illumina). Sequencing was performed using a 150bp paired-end sequencing kit on a MiSeq (Illumina). All stages of the workflow were performed according to the manufacturer's instructions.

Sequence quality: For each sample reported, >95% of the target coding and flanking intronic regions of the 41 genes was covered to a minimum depth of 20 reads (20X). Specific details of coverage and depth for individual tests are available from the laboratory on request.

Variant identification and interpretation: Sequence data were mapped and variants identified using GenomeAnalysisToolKit (GATK) and NextGENe (Softgenetics) with hg19 (GRCh37) human genome as the reference. Variants identified were subsequently classified according to recent ACGS Best Practice Guidelines for Variant Classification using all available evidence. Any clinically significant variants were confirmed by Sanger sequencing.

Variant reporting Variants were reported according to HGVS guidelines using the accession numbers listed below. Variants categorised as non-pathogenic, likely non-pathogenic or cold/cool VUS were not included in the clinical report. Details of these variants are available from the laboratory on request.

Genes included and associated sequence accession numbers:

ACTB (NM_001101.5); **ACTG1** (NM_001614.5); **ALDH1A3** (NM_000693.4); **BCOR** (NM_001123385.2); **C12orf57** (NM_138425.4); **CHD7** (NM_017780.4); **COL4A1** (NM_001845.6); **FOXC1** (NM_001453.3); **FOXE3** (NM_012186.3); **FZD5** (NM_003468.4); **GJA8** (NM_005267.5); **ITPA** (NM_033453.4); **ITPR1** (NM_001378452.1); **MAB21L1** (NM_005584.5); **MAB21L2** (NM_006439.5); **NAA10** (NM_003491.4); **OTX2** (NM_021728.4); **PAX2** (NM_003990.4); **PAX6** (NM_000280.4); **PITX2** (NM_00325.6); **PITX3** (NM_005029.4); **RAB18** (NM_021252.5); **RAB3GAP1** (NM_012233.3); **RAB3GAP2** (NM_012414.4); **RARB** (NM_000965.5); **RAX** (NM_013435.3); **RBP4** (NM_006744.4); **SALL2** (NM_001364564.1); **SALL4** (NM_020436.5); **SHH** (NM_000193.4); **SIX3** (NM_005413.4); **SMCHD1** (NM_015295.3); **SMOC1** (NM_001034852.3); **SOX2** (NM_003106.4); **STRA6** (NM_022369.4); **TBC1D20** (NM_144628.4); **VAX1** (NM_001112704.2); **VSX2** (NM_182894.3); **YAP1** (NM_001130145.3); **ZEB2** (NM_014795.4); **ZIC2** (NM_007129.5).

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