

Supplementary Table 1: Table depicting the ocular tumour-associated genes, their location, function, and associated diseases.

Gene:	Location:	Function:	Disease:
ABCG2	4q22.1	ATP Binding Cassette Subfamily G Member 2	Retinoblastoma
APC	5q22.2	Tumour suppressor gene. Regulator of Wnt Signalling Pathway.	Congenital hypertrophy of retinal pigment epithelium, Gardner syndrome, ocular surface squamous cell neoplasia.
BAP1	3p21.1	Tumour suppressor gene. A deubiquitylase, thought to be a key regulator of cell cycle, growth, response to DNA damage and chromatin dynamics.	Uveal melanoma, conjunctival melanoma
BRAF	7q34	Proto-oncogene. Raf family of serine/threonine kinases. Key regulator MAP kinase/ ERK signalling pathway, affecting cell division, differentiation, and secretion.	Conjunctival melanoma, cutaneous melanoma, uveal melanoma
BRCA2	13q13.1	Tumour suppressor gene. DNA Repair Associated.	Uveal melanoma
CCND1	11q13.3	Proto-oncogene. Regulatory protein in G1/S transition of cell cycle.	Von Hippel-Lindau Syndrome, Uveal melanoma
CDK4	12q14.1	Proto-oncogene. Serine/threonine kinase involved in cell cycle regulation and repair of damage caused by UV radiation.	Uveal melanoma, conjunctival melanoma
CDK6	7q21.2	Proto-oncogene. Serine/threonine kinase involved in cell cycle and differentiation; promotes G1/S transition.	Uveal melanoma, conjunctival melanoma

CDKN2A	9p21.3	Tumour suppressor gene. Capable of inducing cell cycle arrest in G1 and G2 phases.	Uveal melanoma, neurofibromatosis
CTC1	17p13.1	Putative tumour suppressor gene. Component <i>CST</i> complex believed to play a role in promoting DNA replication.	Cerebroretinal microangiopathy with calcifications and cysts
CTNNB1	3p22.1	Proto-oncogene. Key downstream component of canonical Wnt signalling pathway.	Teratocarcinosarcoma of the eye
CYSLTR2	13q14.2	Proto-oncogene. Involved in the production and activity of inflammatory mediators.	Uveal melanoma
DICER1	14q32.13	Haploinsufficient tumour suppressor gene with loss of one allele leading to tumour progression but loss of both alleles having an inhibitory effect on tumour development. Endoribonuclease involved in post-transcriptional gene silencing.	Ciliary body medulloepithelioma, rhabdomyosarcoma
DMPK	19q13.32	Mediator of tumour suppressor genes. Various functions including maintenance of skeletal/ cardiac muscle structure and function, synaptic plasticity and regulation of chloride currents.	Uveal melanoma
DUX4	4q35.2	Tumour suppressor/ proto-oncogene. Transcription factor involved in embryogenesis, physiology & disease.	Coats-like syndrome, exudative retinopathy
EIF1AX	Xp22.12	Proto-oncogene. Encodes an essential eukaryotic	Uveal melanoma

		translation initiation factor. Required for binding of 43S complex to the 5' end of capped RNA.	
ERCC2	19q13.32	DNA repair gene involved in separating the double helix via the 5'-3' helicase activity.	Xeroderma pigmentosum, ocular squamous cell carcinoma
FHIT	3p14.2	Tumour suppressor gene. Important in regulation of apoptosis. Involved in purine metabolism.	Uveal melanoma
FLCN	17p11.2	Tumour suppressor gene. Multi-functional protein, involved in both the cellular response to amino acid availability and in the regulation of glycolysis.	Birt-Hogg-Dubé syndrome, choroidal melanoma, uveal melanoma, lid folliculoma
GNA11	19p13.3	Proto-oncogene. G Protein Subunit Alpha 11	Uveal melanoma, choroidal
GNAQ	9q21.2	Proto-oncogene. G Protein Subunit Alpha Q	Sturge weber syndrome, uveal melanoma, nevus of ota
KDM4C	9p24.1	Regulator of oncogenes and tumour suppressor genes. Nuclear protein involved in histone modification and a regulator of several transcription factors.	Uveal melanoma
KIF11	10q23.33	Proto-oncogene. Protein involved in spindle dynamics during mitosis. Also involved in retinal vascular development.	Familial exudative vitreoretinopathy
LRP5	11q13.2	This gene encodes a transmembrane low-density lipoprotein receptor that binds and internalises ligands in the process of receptor-mediated endocytosis	Osteoporosis-pseudoglioma syndrome

MAPKAPK5	12q24.12-q24.13	Tumour suppressor gene. Serine/threonine kinase involved in cellular stress and inflammatory response.	Uveal melanoma
MC1R	16q24.3	Tumour suppressor gene. Encodes melanocortin one receptor.	Uveal melanoma
MDM2	12q15	Important negative regulator of <i>p53</i> tumour suppressor.	Uveal melanoma, retinoblastoma
MITF	3p13	Tumour suppressor gene. Regulates differentiation and development of melanocytes and retinal pigment epithelium.	Uveal melanoma
MLANA	9p24.1	Protein antigen found on the surface of melanocytes.	Uveal melanoma, conjunctival melanoma, malignant spindle cell melanoma
MLH1	3p22.2	Tumour suppressor gene. Involved in post-replicative DNA mismatch repair.	Keratocanthoma, Muir-Torre syndrome, sebaceous adenoma, cutaneous melanoma
MYC	8p24.21	Proto-oncogene. Multifunctional, nuclear phosphoprotein involved in cell cycle, cell growth, apoptosis, metabolism, biosynthesis, adhesion, and mitochondrial biogenesis.	Uveal melanoma
MSH2	2p21-p16	Tumour suppressor gene. Protein involved in post-replicative DNA mismatch repair system.	Sebaceous adenoma, cutaneous melanoma, uveal melanoma
MYD88	3p22.2	Proto-oncogene. Adapter protein involved in <i>Toll-like</i> receptor and <i>IL-1</i> receptor signalling pathway in innate immune response.	Vitreoretinal lymphoma

NDP	Xp11.3	Activates the canonical Wnt signalling pathway through <i>FZD4</i> and <i>LRP5</i> coreceptor. Involved in retinal vascularisation.	Coats, familial exudative vitreoretinopathy, persistent foetal vasculature, Norrie disease
NF1	17q11.2	Tumour suppressor gene. Appears to be a negative regulator of the ras signal transduction pathway.	Multiple including, Lisch nodules, optic gliomas, capillary haemangiomas
NF2	22q12.2	Tumour suppressor gene. Involved in regulation of contact-dependent inhibition of cell proliferation and functions in cell-cell adhesion and transmembrane signalling.	Hamartomas
NRAS	1p13.2	Proto-oncogene. Regulatory protein in cell division.	Conjunctival melanoma, uveal melanoma, cutaneous melanoma
PDCD1	2q37.3	Tumour suppressor gene. Immune-inhibitory receptor expressed in activated T cells; involved in regulation T cell function.	Conjunctival squamous cell carcinoma, uveal melanoma
PLAG1	8q12.1	Proto-oncogene. Involved in cell proliferation by regulating a wide array of target genes.	Pleomorphic adenoma lacrimal gland
PLCB4	20p12.3-p12.2	Proto-oncogene. Encoding form of <i>phospholipase C</i> involved in <i>phosphoinositide cycle</i> .	Uveal melanoma
POT1	7q31.33	Encodes a protein involved in telomere maintenance.	Uveal melanoma
PRDX3	10q26.11	Mitochondrial protein with antioxidant function.	Uveal melanoma
PTCH1	9q22.32	Tumour suppressor gene. Acts as a receptor for Sonic Hedgehog (<i>SHH</i>).	Ocular surface squamous cell neoplasia, retinoblastoma, basal cell

			carcinoma, Gorlin-Goltz syndrome, orbital teratoma
PTCH2	1q34.1	Tumour suppressor gene. Role in control of cellular growth may be a receptor for <i>SHH</i> .	Gorlin-Goltz syndrome,
PTEN	10q23.31	Tumour suppressor gene. Acts as a dual-specificity protein phosphatase, dephosphorylating tyrosine-, serine- and threonine-phosphorylated proteins.	Cowden syndrome, retinoblastoma, conjunctival melanoma, uveal melanoma, PTEN hamartoma tumour syndrome, choroidal schwannoma
RB1	13q14.2	Tumour suppressor gene. Key regulator of G1/S transition of cell cycle.	Retinoblastoma, ocular sebaceous carcinoma
RBL1	20q11.23	Likely to be a tumour suppressor gene. Key regulator of entry into cell division.	Retinoblastoma
SF3B1	2q33.1	Proto-oncogene. Involved in pre-mRNA splicing as a component of the splicing factor <i>SF3B</i> complex.	Uveal melanoma, conjunctival melanoma
SRSF2	17q25.1	Proto-oncogene. Necessary for the regulation of gene transcription, pre-mRNA splicing, mRNA transport and stability.	Uveal melanoma, iris and ciliary melanocytoma
SUFU	10q24.32	Tumour suppressor gene. Negative regulator in the hedgehog/smoothened signalling pathway.	Retinoblastoma, conjunctival melanoma
TERT	5p15.33	Tumour suppressor gene. Encodes telomerase a ribonucleoprotein enzyme essential for the replication of chromosome termini in most eukaryotes.	Uveal melanoma, conjunctival melanoma

TP53	17p13.1	Tumour suppressor gene implicated in many tumour types; induces growth arrest or apoptosis depending on physiological circumstances and cell type.	Ocular surface squamous cell neoplasia, Li-Fraumeni syndrome, orbital rhabdomyosarcoma, uveal melanoma
TSC1	9q34.13	Tumour suppressor gene. Encodes growth inhibitory protein hamartin.	Tuberous sclerosis
TSC2	16p13.3	Tumour suppressor gene. Encodes growth inhibitory protein tuberlin.	Tuberous sclerosis
U2AF1	21q22.3	Encodes an RNA-binding protein involved in the recognition of the 3' splice site required for recruitment of U2 snRNP during pre-mRNA splicing.	Uveal melanoma
VHL	3p25.3	Tumour suppressor gene. Involved in ubiquitination and subsequent proteasomal degradation via von Hippel-Lindau ubiquitination complex.	Von Hippel-Lindau, Hemangioblastoma
XPA	9q22.33	Involved in DNA excision repair.	Ocular squamous cell carcinoma, pterygium, pinguecula, conjunctival melanoma