MEDGENOME

Ophthatome Knowledgebase: Over 500,000 Clinical Phenotype Records for Ocular Research

Introduction:

Ocular diseases comprise a wide spectrum of diseases affecting any organ of an anterior or posterior segment of the eye, which includes infectious, genetic (monogenic or complex), environmental and trauma. Although infectious diseases are curable by timely intervention, genetic diseases are often untreatable, or poorly managed leading to a high burden of visual impairment (VI) in the population. The most common causes of VI are uncorrected refractive error (43%), cataract (33%), glaucoma (2%), age-related macular degeneration (AMD), diabetic retinopathy, trachoma, corneal opacities - 1% each and 18% with undetermined causes[1]. Advancements in genetic, molecular and proteomics technologies have propelled the ophthalmic research, contributing to a better understanding of various ocular disease pathology, treatment modalities and drugs. Very recently, gene therapy for Leber Congenital Amaurosis (LCA) in patients with RPE65 gene mutation has been approved by the US FDA[2]. This significant clinical development promises to bring more personalized therapies for treating genetic diseases.

While there has been a tremendous progress in understanding the mechanisms of several ocular diseases, complex diseases with a diverse manifestation of phenotypes and sub-phenotypes require studies on larger cohorts with comprehensive longitudinal phenotype and clinical data to address the unmet research needs in this disease space.

To enable genomic, pharmacogenomic and clinical research and discovery for ocular diseases, MedGenome has launched the Ophthatome™ Knowledgebase. This knowledgebase of ocular diseases is a comprehensive collection of clinical, phenotype and biochemical data providing researchers and clinicians with a platform to design studies that address critical unmet needs in eye disorders. The searchable interface allows end users to build complex queries to select disease cohorts based on organs affected, disease type and subtype, the age of disease onset, drug response and many other clinical and phenotypic parameters. In this white paper, we discuss the key features of the database and highlight specific examples of use-cases of the database.

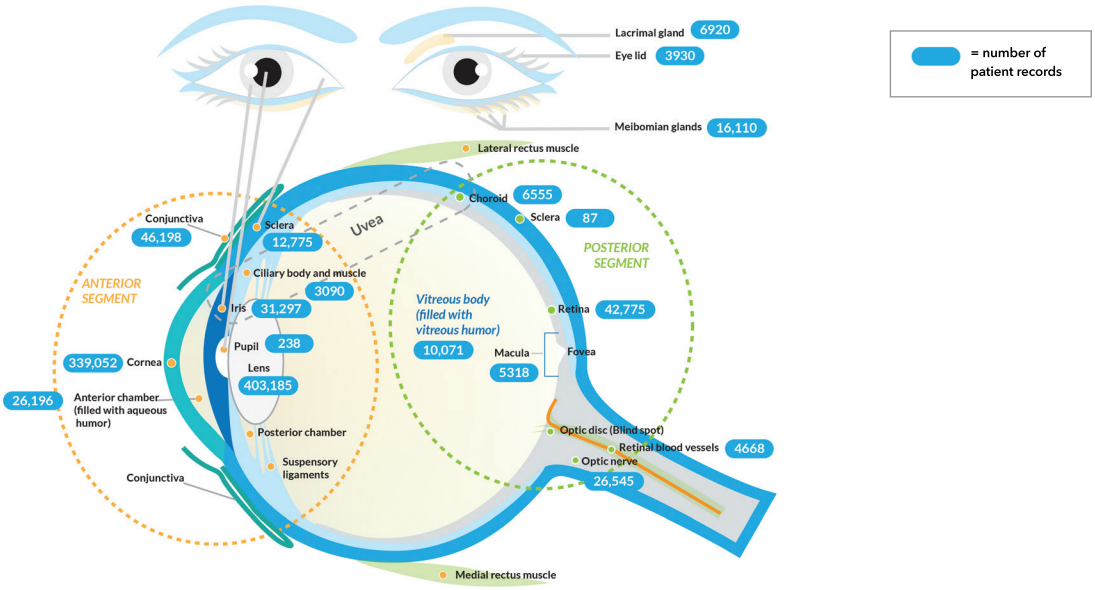

Figure 1: *Overview of the number of patient datasets available for various diseases of different parts of the eye*

Summary of data captured in Ophthatome

- Over 550,000 cases with comprehensive ophthalmic, clinical, biochemical, diagnostic imaging and therapeutic intervention data.
- Over 524 ophthalmic disease types and about 1,800 disease sub-types of 31 different eye parts.
- 40+ clinical variables and ~372,000 images
- Longitudinal view of individuals' clinical profiles.

Figure 2: *Summary of data found in Ophthatome: 2A. Top 25 ocular diseases out of the 524 diseases represented in Ophthatome. 2B: Top 25 ocular diseases affecting individuals over 50 years. 2C) Key clinical data available in Ophthatome for each patient. 2D) Number of subjects with longitudinal data covered over multiple visits to the hospital.*

Representative datasets on selection of disease cohorts using Ophthatome Knowledgebase

Cohorts of specific disease(s) and subtype(s) either alone or in combination with other ophthalmic diseases, additionally, or separately based on disease onset (age-related, congenital etc.), cause (infectious, genetic, developmental, environmental etc.) can be chosen. Cohorts with defined values in parameters like refractive error, intraocular pressure, central corneal thickness, number of visits, different available ophthalmic diagnostic procedural images, systemic disease information can be obtained. These parameters can also be applied on chosen disease type and sub-type thus defining the criteria of the cohort. Lastly, as shown below, there is data on drugs prescribed based on drug category and generics, either alone or in combination with other drugs.

Summary | Cases(1,668) | Filters | Active filters | Search | Save cohort

Visit > 1 | Disease: Diabetic retinopathy (DR) | Drug category : ANTIINFLAMMATORY | Drug category : ANTINEOVASCULARIZATION AGENT | Drug category Opt : Along with | Drug category select : OR | Drugs : both

Id	Age	Sex	Last visit date	Diagnosis	Refraction	Intra ocular pressure	Pachymetry	Glass prescription	Medicine
1225	56 Y	M	06-07-2017	Presbyopia,Regular astigm...	View data	OD:18;OS:14	-	View data	ATORVA TABLET
1630	58 Y	M	20-02-2017	Hypermetropia, right eye,...	View data	OD:7;OS:8	-	View data	NEVANAC (0.1%)
3566	60 Y	F	12-05-2015	Presence of intraocular l...	View data	OD:11;OS:13	-	View data	NEVANAC (0.1%)
6017	58 Y	M	11-05-2017	Type 2 diabetes mellitus ...	View data	OD:12;OS:0	-	View data	DORZOX (2%) E/[

Summary | Cases(118) | Filters | Active filters | Search | Save cohort

Visit > 1 | Disease: Diabetic retinopathy (DR) | Drug category : ANTIINFLAMMATORY | Drug category : ANTINEOVASCULARIZATION AGENT | Drug category Opt : Along with | Drug category select : AND | Drugs : both

Id	Age	Sex	Last visit date	Diagnosis	Refraction	Intra ocular pressure	Pachymetry	Glass prescription	Medicine
6624	63 Y	M	26-06-2017	Myopia, bilateral,Presbyo...	View data	OD:19;OS:12	-	View data	MEGABROM
19139	48 Y	F	07-12-2016	Presbyopia,Regular astigm...	View data	OD:12;OS:0	-	View data	NEVANAC (0
40006	71 Y	M	13-05-2017	SEVERE NPDR,CSME,Central ...	View data	OD:11;OS:8	-	View data	VIGAMOX E/
45816	52 Y	M	27-04-2016	Type 1 diabetes mellitus ...	View data	OD:35;OS:7	-	View data	BRIMOLOL (
51907	60 Y	F	13-06-2017	Age-related nuclear catar...	View data	OD:12;OS:12	-	View data	NEVANAC (0

Figure 3: *Ophthatome interface showing search of patient data for diabetic retinopathy : Of the 14,298 diabetic retinopathy (DR) cases, 1,668 subjects with longitudinal data (i.e.>1 visit) were treated with either Ozurdex OR any one of the anti-vascular endothelial growth factor agents (Bevacizumab, Ranibizumab, Ranibizumab (biosimilar)), 118 were treated with both ozurdex AND any one of the anti-vascular endothelial growth factor agents (Bevacizumab, Ranibizumab, Ranibizumab (biosimilar)).*

Summary:

Ophthatome offers researchers and clinicians a comprehensive choice of large clinically well-defined disease cohorts to investigate disease phenotype, perform statistical analysis on quantitative variables, design methods to investigate genetic and molecular causes to gain insights into the genetics and biology of the diseases and how they impact response to drugs.

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References:

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2. Ameri H. Prospect of retinal gene therapy following commercialization of voretigene neparvovec-rzyl for retinal dystrophy mediated by RPE65 mutation. J Curr Ophthalmol. 2018 Feb 16;30(1):1-2