Glaucoma 1, Open Angle, A

Alternative Names
GLC1A
Glaucoma, Primary Open Angle, Juvenile-Onset, 1
JOAG1
Glaucoma 1, Open Angle, L
GLC1L

Record Category
Disease phenotype

WHO-ICD
Diseases of the eye and adnexa > Glaucoma

Incidence per 100,000 Live Births
Unknown

OMIM Number
137750

Mode of Inheritance
Autosomal dominant; ?distinct from Rieger syndrome

Gene Map Locus
1q24.3-q25.2, 2p22-p21

Description
Glaucoma is a progressive optic neuropathy characterized by a degeneration of the optic nerve, which is usually associated with elevated intraocular pressure. The increase in intraocular pressure is probably caused by a reduction in outflow of aqueous humor through the trabecular outflow pathways. The degeneration of the optic nerve is the result of the loss of individual retinal ganglion cells, where the ganglion cells die by an apoptotic mechanism.

Glaucoma accounts for 15% of blindness worldwide, and is the second leading cause of blindness in the world. It is a heterogeneous group of disorders, the majority of which are associated with an open, normal appearing anterior chamber angle with normal trabecular meshwork and are termed open angle glaucoma. Open angle glaucomas have onset in mid adulthood and relentless slow progression. The juvenile and infantile glaucomas are more severe clinically and more difficult to manage.

Primary open angle glaucoma that develops before the age of 40 years is known as “juvenile-onset primary open angle glaucoma” (JOAG). Autosomal dominant juvenile-onset open-angle glaucoma has a characteristic onset in the second or third decade, high intraocular pressure, poor responsiveness to medical therapy, and frequent dependence on filtering surgery to control both pressure and the attendant optic neuropathy.

Molecular Genetics
Juvenile-onset primary open angle glaucoma can be caused by mutation in the gene encoding myocilin (MYOC). MYOC encodes the protein myocilin, which is believed to have a role in cytoskeletal function, and is expressed in many ocular tissues, including the trabecular meshwork, and was revealed to be the trabecular meshwork glucocorticoid-inducible response protein (TIGR). The MYOC gene is comprised of three exons, and missense mutations associated with juvenile-onset primary open angle glaucoma have been found primarly in the third exon that codes for a protein domain with homology to olfactomedin. The MYOC DNA sequence variants found in patients with glaucoma are associated with a range of disease severity, with some missense mutations causing very severe early-onset disease with autosomal dominant inheritance, whereas other missense mutations and a common truncating mutation (GLN368STOP) are associated with late-onset primary open angle glaucoma.

Epidemiology in the Arab World
Kuwait
Al-Merjan et al. (2005) presented the causes and incidence rates of disorders leading to blindness and low vision in Kuwait, based on the data collected by the Visual Disability Committee in a 5-year period from 2000 to 2004. Of the 826,083 people (407,871 males) registered with blindness and low vision, 39% were below the age of 20-
years, 32% were between the ages of 21 and 40-years, while only about 10% were over 60-years of age. Primary open angle glaucoma was found to occur with an overall incidence rate of 0.24 per 100,000 population. The incidence varied between males (0.44) and females (0.04).

### Oman
Ur'Rahman (1992) conducted a pilot study to estimate the intra ocular pressure in a random Omani population between the ages 40 and 50 years old. He found that 0.5% had definite open angle glaucoma while 2.4% of the cases with an intra ocular pressure more than 21mmHg, had positive provocative test. The author emphasized the need of generalized survey to detect early glaucoma.

Khandekar et al. (2005) estimated the magnitude, components and determinants of noncompliance among glaucoma patients, through a cross-sectional study which included 105 glaucoma patients of Omani nationality (55.2% females and 44.8% males with mean duration of medical treatment for glaucoma of 5.43 years), randomly selected from different regions of Oman. Data on personal details and history of glaucoma, knowledge, beliefs, and the practice of glaucoma treatment was collected by a closed-end questionnaire. There were no significantly different percentages of patients in different age groups and regions, but most of them were illiterate (67.6%) and married (69.5%). Noncompliance information included discontinuation of medications, missing doses, regularity of hospital visits, maintenance of proper intervals between two medications, and the usage of proper methods of installing the drops. Factors that affected compliance (inadequate knowledge, negative attitude, the presence of co-morbidity, complexities of the medication, or improper technique for the installation of eye-drops) were analyzed with estimation of the relative risks and 95% confidence intervals. Noncompliant patients were counseled to improve their glaucoma care. In 24.8%, excellent compliance was detected (mostly with regular follow up and following dosage frequencies), with noncompliance seen in 72.5% (intermediate and poor compliance), which was mostly contributed by discontinuation of medications and not maintaining a proper interval between the medications. It was found that adequate knowledge of glaucoma and its possible complications were negatively associated with noncompliance, while no association was determined among glaucoma patients between a positive attitude and proper practice and noncompliance. Upon analyzing compliance in relation to age, gender, region of residence, educational level, marital status, and duration of treatment, no association was detected. Khandekar et al. (2005) recommended further longitudinal studies to further estimate the causes of noncompliance with glaucoma treatment in Oman, and in order to improve compliance and reduce visual impairment, they advised on improving knowledge, attitude, and practice, while making the drug regimen patient friendly.

### Qatar
Al-Mansouri (2002) studied a sample of patients with primary glaucoma to determine the pattern, severity, and medical risk factors associated with the condition in Qatar. A sample of affected Qatari nationals, aged over 30 years, was interviewed by means of a questionnaire as well as clinical examinations. The sample consisted of 195 patients with 352 glaucomatous eyes. Of these, 137 patients (58.4% females) were affected with POAG. There was a positive family history in 34.3% of patients with POAG and 34.5% of patients with Primary Angle Closure Glaucoma (PACG). This study population showed a surprisingly early onset of glaucoma. About 36% of patients presented with POAG in the first 50 years of life, and 21% before 40-years of age. In addition, a significant number of patients were legally blind due to glaucoma at the time of their first presentation. A total of 55 eyes with POAG had visual acuity between 6/60 and no perception of light, and 36.4% of the patients had advanced glaucomatous optic disc cupping. Compliance with administration of medical therapy was low in this population, with 47.7% of the whole series showing poor compliance. About 65% of this group showed progressive glaucoma changes. Al-Mansouri (2002) recommended the need for wide-scale health education, screening, and early detection of glaucoma in the Qatari population.

### Saudi Arabia
Andersen et al. (1996) used six polymorphic DNA markers on chromosome 1q, in a region showing tight linkage to GLC1A, in 25 Saudi Arabian families showing PCG. Four of the families demonstrated the presence of two regions of exclusion, which spanned the entire length of the 8cM region containing the GLC1A locus. In the remaining 21 families, no PCG locus was shown to segregate in an autosomal recessive manner on haplotypes constructed using markers in this region. The results of the study, therefore, revealed the exclusion of the 8cM region in each of these families.

### References


Related CTGA Records
Cytochrome P450, Subfamily I, Polypeptide 1
Glaucoma 3, Primary Infantile, A

External Links
http://www.emedicine.com/oph/topic333.htm
http://www.glaucoma.org/learn/what_is_glaucoma.html
http://www.orpha.net/data/patho/GB/uk-glaucoma.pdf

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