Exploring Social and Genetic Factors in Eye Health Disparities

Darani Ashley Thammavongsa, BS, MS, PA-S[1]
Sean Kolhoff, PhD [2]
Melissa Murfin, PhD, PA-C[1]
[1]Ithaca College, [2]American Academy of Physician Assistants

Abstract

This study examines the relationship between social determinants of health and genetic predisposition on the prevalence and co-occurrence of chronic eye conditions such as cataracts, age-related macular degeneration, glaucoma, and diabetic retinopathy among participants in the *All of Us* Research Program. It highlights the impact of socioeconomic factors and genetic predispositions on these conditions, informing healthcare strategies to better serve diverse populations.

Introduction

Eye diseases such as diabetic retinopathy, cataracts, age-related macular degeneration (AMD), and glaucoma are major causes of vision loss in the United States. While genetic predisposition is a well-known risk factor, growing evidence suggests that social determinants of health—including insurance status, income, employment, and health behaviors like smoking—also contribute significantly to disease risk. Understanding the combined impact of genetic and socioeconomic factors is essential for developing comprehensive screening and prevention strategies.

Methods

A retrospective analysis was conducted using summary-level data from the *All of Us* Research Program. Participants were categorized by the presence of one or more chronic eye conditions, including diabetic retinopathy, cataracts, age-related macular degeneration (AMD), and glaucoma. Family history and social determinants—such as health insurance status, income, employment, smoking, alcohol use, and neighborhood safety—were examined. Odds ratios and 95% confidence intervals were calculated using grouped logistic regression models to evaluate the association between these predictors and eye disease prevalence.

Figure 1.

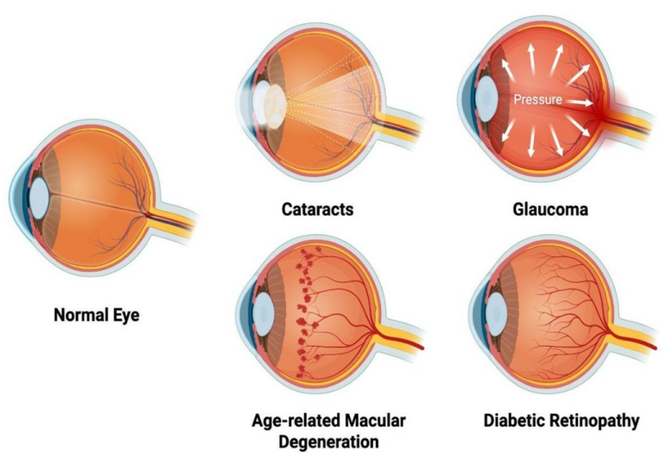

Figure 1. Comparison of a Normal Eye to Four Common Ocular Conditions
Cataracts cause clouding of the lens, impairing light transmission.
Glaucoma involves increased intraocular pressure that damages the optic nerve.
Age-related macular degeneration leads to central vision loss due to retinal degeneration.
Diabetic retinopathy results from retinal blood vessel damage caused by prolonged hyperglycemia.
Image created with BioRender.com.

Table 1.

Demographics of Participants with Eye Conditions (Total N = 409,420)							
Category	Group	Cataracts	Glaucoma	Age-Related Macular Degeneration	Diabetic Retinopathy	n (with eye condition)	% of Total Population
Age	18-49	94	2,073	177	4,425	6,729	1.64
	50-89	3,098	16,823	5,276	23,132	48,329	11.80
	>89	28	224	199	310	761	0.19
Sex (Assigned at Birth)	Female	1,240	3,814	1,103	5,298	11,455	2.80
	Male	427	1,818	568	2,649	5,462	1.33
	Other	41	144	40	194	419	0.10
Race	White	1,503	4,229	1,409	6,290	13,431	3.28
	Black	60	636	55	606	1,357	0.33
	Asian	27	216	44	207	494	0.12
	Native Hawaiian or Pacific Islander	<20	<20	<20	<20	80	0.02
	Middle Eastern	<20	<20	<20	30	90	0.02
	Multi-Racial or Other Race	123	669	179	985	1,936	0.48
Ethnicity	Hispanic	24	398	105	613	1,140	0.28
	Non-Hispanic	1,599	5,101	1,511	7,156	15,367	3.75
	Other	85	277	95	372	829	0.20
Total with eye condition(s)		3,220	19,120	5,652	27,867	55,859	13.64

Table 1. Participant Demographics by Eye Condition
Data from the *All of Us* Research Program (N = 409,420) show the distribution of Cataracts, Glaucoma, ARMD, and DR across key demographic groups. Most eye condition cases occurred in participants aged 50–89. Females, White, and Non-Hispanic individuals accounted for the largest proportions. Each row shows the number of affected participants (n) and their percentage of the total cohort.

Table 2.

Social Determinants of Health Among Participants with Eye Conditions (Total N = 409,420)							
Category	Group	Cataracts	Glaucoma	Age-Related Macular Degeneration	Diabetic Retinopathy	n (with eye condition)	% of Total Population
Health insurance status	Yes	3,100	18,162	5,413	26,362	53,037	12.95
	No	23	436	76	686	1,221	0.30
Neighborhood safety	Agree	1,592	5,739	1,961	8,288	17,580	4.29
	Disagree	134	523	160	797	1,614	0.39
Household income	0-50k	904	7,123	1,836	10,268	20,131	4.92
	50k-100k	841	3,740	1,222	5,360	11,163	2.73
	100k >	1,059	3,769	1,269	5,713	11,810	2.89
Employment status	Employed	925	5,824	1,304	9,007	17,060	4.17
	Retired	2,009	9,183	3,710	12,494	27,396	6.69
	Not Employed or Other	530	3,765	941	8,497	13,713	3.34
Smoking Frequency	Yes	69	275	102	454	900	0.22
	No or Skip	2,174	6,947	2,262	9,700	21,063	5.15
Alcohol Drink Frequency	Weekly (>2 drinks per week)	881	2,585	940	3,505	7,911	1.93
	Monthly (<4 drinks per month)	794	2,444	753	3,604	7,595	1.85
	Never or Skip	573	2,193	675	3,045	6,446	1.58

Table 2. Participant Social Determinants of Health by Eye Condition
Social determinants of health among participants with eye conditions (N = 409,420). Table displays the number and percentage of individuals diagnosed with cataracts, glaucoma, ARMD, and diabetic retinopathy across key socio-demographic groups.

Figure 2.

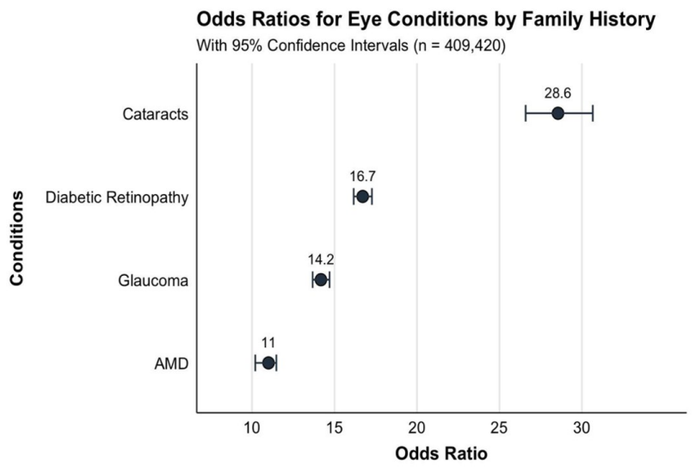

Figure 2. Odds Ratios for Eye Conditions by Family History
Odds ratios (OR) with 95% confidence intervals are shown for participants reporting a family history of Cataracts, Diabetic Retinopathy, Glaucoma, or Age-Related Macular Degeneration (AMD) in the *All of Us* Research Program (N = 409,420). A family history was most strongly associated with Cataracts (OR = 28.6), followed by Diabetic Retinopathy (OR = 16.7), Glaucoma (OR = 14.2), and AMD (OR = 11.0).

Results

This analysis examined the relationship between family history, social determinants of health, and the risk of diabetic retinopathy, cataracts, age-related macular degeneration (AMD), and glaucoma. Family history was significantly associated with increased risk for all conditions, with odds ratios ranging from approximately 11.0 (AMD) to 28.6 (cataracts), and all 95% confidence intervals above 1. Among social factors, diabetic retinopathy was more prevalent among uninsured individuals (686 cases in 1,221) compared to insured (26,362 in 53,037), and AMD was most common in those with incomes below $50,000 (3,353 cases). Current smokers showed elevated rates of cataracts (1,635) and diabetic retinopathy (9,106), while alcohol use was widespread but less associated. These findings support the need to integrate both genetic and social risk factors into eye disease screening and prevention strategies.

Conclusion

The study emphasizes family history and social determinants such as insurance status, income, employment, and smoking were significantly associated with increased risk for major eye conditions. These results highlight the importance of incorporating both genetic and socioeconomic factors into eye disease screening and prevention strategies.

Discussion

Genetic predispositions significantly influence the risk of various eye conditions, highlighting the importance of family history in clinical assessments.

- **Limitations:** As a retrospective analysis using observational data, this study is subject to potential biases such as misclassification, missing data, and unmeasured confounders, which may affect the accuracy of associations observed.
- **Clinical Implications:** Detailed family histories are crucial for early detection and intervention strategies, potentially improving patient outcomes.
- **Social Determinants:** Factors such as insurance status, income, employment, and smoking behavior also showed strong associations with eye disease risk, underscoring the need for a holistic approach to screening and prevention.
- **Future Research:** Future studies should apply GWAS to identify loci linked to AMD, glaucoma, cataracts, and diabetic retinopathy, enhancing genetic risk profiling.

References and Acknowledgements

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