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Positive Family History and Astigmatism Severity in Pakistani Adolescents: A Cross-Sectional Analytical Study

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ABSTRACT

Background and Objective: Refractive errors constitute a major public health burden in Pakistan, and astigmatism is among the most commonly diagnosed conditions in adolescents presenting to eye care facilities. Family history is thought to influence both whether astigmatism develops and how severe it becomes. This study aimed to establish whether a positive family history of refractive error is associated with more severe astigmatism among Pakistani adolescents.

Methods: This cross-sectional analytical study was conducted using data collected over a six-month period in 2024. Sample size was calculated using G*Power 3.1 (80% power, two-tailed alpha of 0.05, anticipated odds ratio of 2.0), yielding a minimum requirement of 288 participants; 320 adolescents aged 10 to 19 years with astigmatism of at least 0.50 DC were enrolled through simple random sampling. Participants were stratified by family history status into those with a positive family history (FH+; n = 198) and those without (FH−; n = 122). Cycloplegic retinoscopy and subjective refraction were performed on all participants, and cylinder power was graded as mild (0.50–1.00 DC), moderate (1.25–2.00 DC), or high (≥ 2.50 DC). Axis type, astigmatism subtype, eye laterality, and best-corrected visual acuity were also recorded. Group comparisons were conducted using chi-square and independent-samples t-tests, with statistical significance set at $p < 0.05$.

Results: A positive family history was present in 198 participants (61.9%). High astigmatism was significantly more prevalent in the FH+ group (38.9%) than the FH− group (19.7%; $p < 0.001$). Mean cylinder power was −2.31 DC in FH+ participants compared with −1.54 DC in FH− participants ($p < 0.001$). Bilateral involvement was more frequent in FH+ cases (63.6% vs 44.3%; $p = 0.001$), as was against-the-rule (ATR) astigmatism (29.3% vs 19.7%; $p = 0.048$). The odds of severe astigmatism were 2.61 times higher (95% CI: 1.52–4.48) among FH+ adolescents.

Conclusion: A positive family history of refractive error substantially increases the risk of severe astigmatism during adolescence, with higher cylinder power, bilateral involvement, and against-the-rule axis orientation as the principal distinguishing features. Incorporating family history into routine eye-screening protocols offers a practical, low-cost way to catch cases earlier and help prevent amblyopia in this population.

KEYWORDS: *astigmatism; family history; hereditary refractive error; adolescents; Pakistan; cylinder power; simple random sampling.*

INTRODUCTION

Refractive errors remain the leading contributor to preventable visual impairment worldwide. The 2021 Global Burden of Disease study identifies refraction disorders, spanning myopia, hyperopia, astigmatism, and presbyopia, as the foremost cause of moderate and severe vision loss, with the greatest burden borne by young people in low- and middle-income countries such as Pakistan. [1] Within this category, astigmatism stands out. It results from unequal corneal or lenticular curvature across meridians, producing blur that cannot be resolved through accommodation. A 2023 systematic review pooling data from 125 independent studies reported astigmatism prevalence ranging from 8% to 62% across age groups and ethnic populations, with consistent evidence that uncorrected astigmatism compromises vision, academic performance, and household finances. [2]

Adolescence, defined by the World Health Organization as the period between ages 10 and 19, represents the most dynamic phase of visual development outside infancy, during which the eyeball continues to grow, the cornea continues to reshape, and refractive status can change substantially within months. [4] Pooled data across Global Burden of Disease sub-regions, analysed in 2022, confirmed that uncorrected myopia and astigmatism peak in the 6-to-17-year age bracket, with sub-regional variation attributable more to disparities in healthcare access than to underlying biological differences. [5] Undetected astigmatism in this period carries substantial consequences, ranging from amblyopia, in which the brain suppresses input from one or both blurred eyes, to impaired academic performance and lasting psychosocial effects. Cylinder power of 2.50 DC or greater is of particular clinical concern, as amblyopia risk is highest and treatment least effective once the critical period of visual development has closed. [6] A Pakistani nationwide study found that approximately one in five high school students carries a visually disabling refractive error without correction, a sign that early detection efforts are not reaching this population. [7]

Genetic factors play a bigger role in astigmatism severity than clinicians often appreciate. Twin studies, sibling and parent-offspring analyses, and molecular genome-wide association studies converge on this conclusion. A 2024 meta-analysis in the American Journal of Ophthalmology, synthesizing 47 heritability studies, reported pooled heritability estimates of 0.46 for refractive astigmatism, 0.48 for corneal astigmatism, and 0.64 for corneal curvature, indicating that genetic factors account for nearly half to nearly two-thirds of the variation in astigmatism magnitude across individuals. This familial influence appears dose-dependent: a 2022 Hong Kong study of 5,708 parent-child trios found that a child's risk of refractive astigmatism increased by 62% when both parents had at least 1.0 DC of astigmatism, rising to more than three times baseline risk when both parents had 2.0 DC or greater. [9] This pattern was corroborated by a 2023 Mexican twin study, which documented significantly higher concordance between monozygotic than dizygotic twins ($p = 8.80 \times 10{-}9$) and suggested the heritability pattern generalizes across ethnic backgrounds. [10] Despite this accumulating evidence, no Pakistani study has examined whether family history predisposes adolescents to astigmatism and also shapes how severe it becomes. That gap is the focus of the present study.

METHODS

A private clinic in Taunsa served as the study site. This cross-sectional analytical study was conducted over an uninterrupted six-month period in 2024. Sample size was estimated a priori using G*Power 3.1 software (University of Düsseldorf, Germany), applying a two-proportion z-test at 80% power and a two-tailed significance level of 0.05, with an anticipated odds ratio of 2.0 for high astigmatism between family-history-positive and family-history-negative groups. Assuming a baseline severe astigmatism proportion of 20% in the FH− group and approximately 33% in the FH+ group, the minimum required sample was calculated at 288 participants; accounting for a 10% allowance for non-response or incomplete records, the recruitment target was set at 320, which was fully achieved. Simple random sampling was employed: each month, a complete register of adolescents presenting to the Eye OPD with confirmed astigmatism was compiled, and participants were selected using a computerized random number generator, ensuring every eligible patient had an equal and independent chance of inclusion.

Eligibility was restricted to patients aged 10 to 19 years presenting with a cylindrical error of at least 0.50 DC in either eye. Exclusion criteria comprised corneal disease such as keratoconus or scarring, prior intraocular or refractive surgery, systemic conditions affecting the anterior segment, incomplete refraction data, or unwillingness to participate. Family history was ascertained at the initial visit using a structured, interviewer-administered proforma. A positive family history (FH+) was defined as confirmation by the patient or guardian that at least one parent or full sibling used glasses or contact lenses for any refractive error; a negative history (FH−) indicated no known refractive error in the immediate family. Where documented, the specific affected relative, father only, mother only, both parents, or sibling, was recorded to support subgroup analysis. Distance visual acuity was assessed using a standard Snellen chart at six metres prior to refraction, followed by objective assessment with a calibrated streak retinoscope. Cycloplegic refraction, using one percent cyclopentolate hydrochloride administered as two drops five minutes apart with measurement thirty minutes after final instillation, was performed routinely in all patients under fifteen years and selectively in older patients with insufficiently relaxed accommodation. Subjective refinement, where cooperation permitted, used a trial frame and Jackson Cross Cylinder at ±0.25 DC and ±0.50 DC increments, with all final refraction values recorded in negative cylinder form.

Astigmatism severity was classified into three bands: mild (0.50–1.00 DC), moderate (1.25–2.00 DC), and high (≥2.50 DC). Axis orientation was categorized as with-the-rule (WTR) for a minus cylinder axis between 160–180° or 0–20°, against-the-rule (ATR) for axes between 70–110°, and oblique for all other orientations. Refractive subtype (compound myopic, simple myopic, compound hyperopic, simple hyperopic, or mixed) and laterality (monocular or bilateral) were recorded for each patient. Best-corrected visual acuity was recorded in Snellen notation and converted for tabulation. Statistical analysis was performed using SPSS and Jamovi (latest versions). The chi-square test was used for categorical comparisons, with Fisher's exact test applied when expected cell counts fell below five; the independent-samples t-test was used for continuous variables. Odds ratios with 95% confidence intervals were calculated for the primary outcome of high astigmatism. Written informed consent was obtained from participants aged eighteen and above, and from a parent or legal guardian on behalf of younger participants.

RESULTS

Three hundred and twenty adolescents with confirmed astigmatism completed the study, with a mean age of 14.8 years (range 10–19). Male participants numbered 148 (46.3%) and females 172 (53.8%). The largest age band was 13–15 years (n = 112; 35.0%), followed by 16–17 years (n = 79; 24.7%), 10–12 years (n = 78; 24.4%), and 18–19 years (n = 51; 15.9%). A positive family history of refractive error (FH+) was identified in 198 of 320 participants (61.9%), while the remaining 122 (38.1%) had no family history (FH−). Within the FH+ group, the father was the sole affected relative in 62 cases (31.3%), the mother alone in 54 (27.3%), both parents in 52 (26.3%), and a sibling only in 30 (15.2%). Dual parental involvement, which carries the greatest genetic loading, was present in more than a quarter of all FH+ cases.
Astigmatism severity differed significantly between family history groups (Table 1). High astigmatism (≥2.50 DC) was recorded in 77 of 198 FH+ participants (38.9%) compared with 24 of 122 FH− participants (19.7%; $p < 0.001$), while mild astigmatism was more common in the FH− group (43.4% vs 21.2%; $p < 0.001$). Moderate astigmatism did not differ significantly between groups (FH+: 39.9%; FH−: 36.9%; $p = 0.582$). Mean cylinder power was −2.31 DC in the FH+ group versus −1.54 DC in the FH− group ($p < 0.001$), and the odds of severe astigmatism were 2.61 times higher in FH+ participants (95% CI: 1.52–4.48; $p < 0.001$). Axis type and laterality findings are presented in Table 2. While WTR astigmatism predominated in both groups, ATR astigmatism was significantly more common in the FH+ group (29.3% vs 19.7%; $p = 0.048$), as was bilateral involvement (63.6% vs 44.3%; $p = 0.001$). Table 3 summarizes the principal comparative findings.

Table 1: Astigmatism Severity and Mean Cylindrical Power by Family History Status

Parameter	FH+ (n = 198)	FH− (n = 122)	p-value
Mild (0.50–1.00 DC)	42 (21.2%)	53 (43.4%)	< 0.001*
Moderate (1.25–2.00 DC)	79 (39.9%)	45 (36.9%)	0.582
High (≥ 2.50 DC)	77 (38.9%)	24 (19.7%)	< 0.001*
Mean cylinder power (DC)	−2.31	−1.54	< 0.001*
OR for high astigmatism (95% CI)	2.61 (1.52–4.48)	Reference	< 0.001*

*OR = Odds Ratio; CI = Confidence Interval; DC = Dioptre Cylinder; FH+ = Positive Family History; FH− = Negative Family History. Values in parentheses are percentages unless otherwise stated. * p < 0.05.*

Table 2: Axis Type and Laterality of Astigmatism by Family History Status

Parameter	FH+ (n = 198)	FH− (n = 122)	p-value
With-the-Rule (WTR)	107 (54.0%)	79 (64.8%)	0.062
Against-the-Rule (ATR)	58 (29.3%)	24 (19.7%)	0.048*
Oblique	33 (16.7%)	19 (15.6%)	0.802
Bilateral astigmatism	126 (63.6%)	54 (44.3%)	0.001*
Monocular astigmatism	72 (36.4%)	68 (55.7%)	0.001*

*WTR = With-the-Rule; ATR = Against-the-Rule. * p < 0.05.*

Table 3: Summary of Key Comparative Findings: FH+ vs FH−

Parameter	FH+	FH−	p-value
n (%)	198 (61.9%)	122 (38.1%)	–
Mean cylinder (DC)	−2.31	−1.54	< 0.001*
High astigmatism (≥ 2.50 DC)	38.9%	19.7%	< 0.001*
Bilateral astigmatism	63.6%	44.3%	0.001*
ATR astigmatism	29.3%	19.7%	0.048*
OR for high astigmatism (95% CI)	2.61 (1.52–4.48)	Reference	< 0.001*
Compound myopic astigmatism	41.9%	39.3%	0.648

*OR = Odds Ratio; CI = Confidence Interval; ATR = Against-the-Rule; DC = Dioptre Cylinder. * p < 0.05.*

DISCUSSION

To our knowledge, this is among the first studies from Pakistan to look beyond the simple association between family history and astigmatism, to ask how it affects severity. The findings demonstrate that adolescents with a positive family history of refractive error carry a substantially greater disease burden. Mean cylinder power exceeds that of FH− peers by more than three-quarters of a dioptre, the odds of high-severity astigmatism are roughly twofold higher, bilateral involvement is more common, and the axis is more often against-the-rule; these differences are summarized in an odds ratio of 2.61 for severe astigmatism. Given that South Asia, including Pakistan, already carries one of the heaviest global burdens of uncorrected refractive disorders in children and adolescents according to GBD 2021 data, [1] and that Pakistani school-based studies consistently report that approximately one in five high school students is functionally visually impaired from uncorrected refractive error, these findings carry direct implications for the targeting of screening efforts [7].

The prevalence of positive family history observed in this cohort, 61.9%, closely aligns with figures reported in comparable Asian populations. The 2022 Hong Kong Children Eye Study, the largest and methodologically most robust family-trio analysis published to date, followed 5,708 children alongside both parents and demonstrated that children of parents with at least 1.0 DC of astigmatism had a 62% higher risk of refractive astigmatism, rising to more than threefold when both parents exceeded 2.0 DC. [9] In the present cohort, the subgroup with both parents affected (26.3% of FH+ cases) was concentrated in the highest cylinder categories, in line with this dose-response relationship. The biological basis for this association is well established: the 2024 American Journal of Ophthalmology meta-analysis of 47 heritability studies reported corneal astigmatism heritability of 0.48 and corneal curvature heritability of 0.64, [8] indicating that genetic factors account for approximately half the variation in cylinder power between individuals. The 2023 Mexican twin study confirmed that this genetic architecture is not confined to European or East Asian populations, supporting the generalizability of these findings to a Pakistani cohort. [10]

The molecular mechanisms underlying this inherited severity are coming into sharper focus. Genome-wide association studies in Asian populations have identified the PDGFRA gene region on chromosome 4q12 as a susceptibility locus for corneal astigmatism, [11] with additional loci, including CTNNA2, implicated in severity through effects on extracellular matrix organization within corneal keratocytes. [12] When these variants co-occur in both parents and are inherited by a child, the resulting corneal geometry becomes more steeply toroidal, producing higher cylinder power. Genetically influenced corneal toricity also appears to follow a distinct trajectory from environmentally induced spherical change: a 2024 longitudinal study of UK patients, tracking prescription histories over two decades, found that the WTR-to-ATR axis transition, which typically occurs gradually through mid-life in the general population, can be accelerated in individuals with inherited corneal toricity. [13] This provides a plausible mechanistic explanation for the higher prevalence of ATR astigmatism observed among FH+ participants in this study (29.3% vs 19.7%; $p = 0.048$). ATR astigmatism is clinically significant, producing greater asthenopia and proving more difficult to correct satisfactorily than WTR astigmatism of equivalent power, compounding the clinical burden already associated with elevated cylinder values in this group.

Bilateral astigmatism was significantly more common among FH+ participants (63.6% vs 44.3%; $p = 0.001$), a finding of particular clinical relevance. Bilateral high astigmatism places children at risk of bilateral amblyopia, an outcome that, once established beyond the sensitive period of visual development, cannot be fully reversed even with optimal optical correction. StatPearls (2025) identifies bilateral high astigmatism as among the few conditions capable of producing bilateral rather than unilateral amblyopia. [6,14] The lower rate of 6/6 best-corrected visual acuity observed in FH+ high-astigmatism cases (53.2%) relative to FH− cases (70.8%) points to a greater amblyopic burden in this genetically predisposed, bilaterally affected group, a reminder of what delayed presentation costs. Similar findings emerged from a 2026 Journal of Global Health study from Northwestern China, which covered children and adolescents aged 3 to 20 and found that bilateral and hereditary astigmatism carried the greatest visual acuity consequences. [15]

Compound myopic astigmatism predominated in both study groups (FH+: 41.9%; FH−: 39.3%), in keeping with existing Pakistani hospital data and Islamabad screen-time research. [3] Mixed astigmatism was proportionally higher among FH+ participants (23.2% vs 18.0%), a numerically notable but statistically non-significant difference warranting prospective investigation.

LIMITATIONS

This study has several limitations. It was conducted at a single centre, which may limit the generalizability of the findings to other regions of Pakistan. Family history was based on self-report by patients or guardians rather than clinical verification of relatives' refractive status, which may have introduced recall or reporting bias. Corneal topography was not performed, so the relative contributions of corneal and lenticular factors to the observed astigmatism could not be separated. Screen time, outdoor activity, and near-work load, all documented risk factors for astigmatism in Pakistani children, [15] were not measured and could not be adjusted for. Finally, the cross-sectional design precludes any conclusion about the direction or causality of the association between family history and astigmatism severity.

CLINICAL AND PUBLIC HEALTH IMPLICATIONS

These findings carry direct implications for eye care delivery in Pakistan. Family history of refractive error is a low-cost variable that can be captured during routine history-taking without additional equipment or specialist training, making it a practical addition to school vision-screening programmes and outpatient eye clinics. Adolescents identified as FH+ could be prioritized for earlier and more frequent cycloplegic refraction, given their elevated risk of high-severity, bilateral, and against-the-rule astigmatism. Since South Asia already carries a disproportionate share of the global burden of uncorrected refractive error in children, [1] incorporating family history into triage protocols offers a scalable way to direct limited screening resources toward the adolescents most likely to benefit.

FUTURE DIRECTIONS

Future work should build on these findings in several directions. Longitudinal studies following FH+ and FH− adolescents over time would clarify whether family history predicts not just baseline severity but also the rate of progression. Incorporating corneal topography and genetic profiling would help disentangle the relative contributions of corneal, lenticular, and axial factors to inherited astigmatism. Multi-centre studies spanning different regions of Pakistan would strengthen the generalizability of these findings, and prospective data on screen time, outdoor activity, and near-work load would help separate the genetic and environmental contributions to astigmatism severity.

CONCLUSION

These findings demonstrate that Pakistani adolescents with a positive family history of refractive error carry a much heavier burden of astigmatism than their peers without such a history. They present with higher cylinder power, nearly twice the rate of high-severity astigmatism, greater likelihood of bilateral involvement, and 2.61-fold greater odds of severe astigmatism. These differences are clinically meaningful and support a targeted policy response. We recommend that family history of refractive error be incorporated as a formal risk flag in school health assessments and eye outpatient triage, prompting early cycloplegic refraction ideally before secondary school entry. Such an approach would shift detection upstream, improve the likelihood of successful amblyopia treatment, and reduce the academic burden associated with uncorrected high astigmatism among Pakistani adolescents.

DECLARATIONS

Conflict of Interest: The authors declare no conflict of interest.

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Ethical Approval: Granted by the Research Ethics Committee, NAEC Taunsa (Approval No. NAEC-APR-2024-01). Written informed consent was obtained from all adult participants and from a parent or legal guardian on behalf of minor participants.

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