© AUG 2026 | IRE Journals | Volume 10 Issue 2 | ISSN: 2456-8880
DOI: https://doi.org/10.64388/IREV10I2-1722223

Personalised Dry Eye Treatment: The Use of Genetic Testing to Tailor Treatment Plans to Individual Patients

DR. Y.R. ERONINI[1], N.C. IKORO[2], A.U. MEGWAS[3], DR. Y.C. AZUAMAH[4], DR. C.M. CHUKWUOHA[5], DR. G. UGWUOKE[6]
[1,2,3,4,5,6]*Federal University of Technology, Owerri.*

Abstract- ***Dry eye disease is a chronic condition affecting millions of people worldwide, causing discomfort, visual impairment, and decreased quality of life. This research was designed as a clinical and laboratory based study for investigating the use of genetic testing to tailor treatment plans to individual patients. One hundred and twenty subjects, 58 (48.3%) males and 62 (51.7%) females with dry eye disease between the ages of 18-65 years were enrolled in the study. Tear assessment, Tear Break Up Time test (TBUT) using fluoresciene dye and tear flow rate test using schirmer strip without anesthesia were done clinically to confirm Dry Eye Disease (DED), tear film samples were collected using sterile swab and genetic investigations were carried out in the molecular laboratory to detect the enrichment of variants responsible for Dry Eye Disease (DED) and Ocular Surface Inflammation (OSI); DNAJB6 was detected in 59 subjects and absent in 61 subjects, MAML3 was detected in 47 and absent in 73 subjects, LINC02267 was detected in 13 subjects and absent in 107 subjects, MUC16 was detected in 14 subjects and absent in 106 subjects, SIRPB3P was detected in 48 subjects and absent in 72 subjects, GAS2L3 was detected in 13 subjects and absent in 107 subjects, KLF4 was detected in 81 subjects and absent in 89 subjects, and PAX6 was detected in 28 subjects and absent in 92 subjects. LINCO2267 and GAS2L3 were the least detected among genetic variants for OSI and DED respectively while KLF4 was the most detected among genetic variants for OSI and DED. The significant representation of DNAJB6, MAML3, LINC02267, and MUC16 variants shows molecular relationship between DED and OSI. From SPSS version 23 data output, data analysis using the Pearson Product Moment Correlation Coefficient at 0.05 level of significance revealed for DED genetic variants a p-value of 0.00. Since p(0.00) <0.05, there is an association between genetic variants and dry eye disease. For ocular surface inflammation, SPSS version 23 data output, data analysis using the Pearson Product Moment Correlation Coefficient at 0.05 level of significance revealed a p-value of 0.00. Since p(0.00) < 0.05, there is an association between genetic variants and ocular surface inflammation. In conclusion, the results of the study showed that dry eye disease and ocular surface inflammation were strongly influenced by genetic variation in epithelial and inflammatory pathways. The results also showed that the initiation and progression of dry eye were multifactorial. It is therefore recommended that eye care practitioners consider genetic testing as an alternative approach for the diagnosis and treatment of dry eye disease. Incorporating genetic testing alongside regular eye examinations can help eye care practitioners better understand each patient's condition.***

Keywords: ***Assessment, Impairment, Investigations, Inflammation, Multifactorial.***

I. INTRODUCTION

Dry eyes, also known as dry eye syndrome (DES), dry eye disease (DED), ocular surface disease (OSD), dysfunctional tear syndrome (DTS), and keratoconjunctivitis sicca (KCS), are among the most common reasons for a visit to an eye doctor (Huang et al., 2022). The definition of a dry eye according to the Tear Film and Ocular Surface Society (TFOS) Dry Eye Workshop II (DEWS II) is, Dry eye is a multifactorial disease of the ocular surface characterized by a loss of homeostasis of the tear film and accompanied by ocular symptoms, in which tear film instability and hyperosmolarity, ocular surface inflammation and neurosensory abnormalities play etiologic roles (Craig et al., 2017).

Genetics is the study of heredity, and seeks to explain the mechanism of hereditary transmission, as well as the genetic basis of individual variation. Medical genetics is the science of genetically associated biologic variation relevant to human traits and diseases. In industrialized countries, improvements in public health and medical care have resulted in a marked reduction in mortality from infectious causes, or nutritional deficiencies; during this time, there has

been a corresponding increase in awareness of the role of genetic factors in human diseases, including neurological disease (Armstrong, Shipkova, Von-Ahsen, & Oellerich, 2004).

Dry eye syndrome (DES), also referred to as dry eye disease (DED) or keratoconjunctivitis sicca (KCS), encompasses multifactorial ocular surface pathology causing discomfort and visual disturbances. Understanding the complexity of tear film composition and dysfunction is pivotal in assessing subjects presenting with dry eyes. This activity delves into the tear film's structure, comprising lipid, aqueous, and mucin layers, impacting tear stability and ocular surface health. It navigates the nomenclature nuances, differentiating between DES and DED, acknowledging their broader clinical implications and comprehensive categorizations (Chen et al., 2010).

Potential causes or factors associated with Dry Eye
Systemic medications: Antihistamines, Antihypertensive, Anxiolytics/benzodiazepines, Diuretics, Systemic hormones, Nonsteroidal antiinflammatory drugs, Systemic or inhaled corticosteroids, Anticholinergic medications, Isotretinoin (causes meibomian gland atrophy), Antidepressants (Paulsen et al., 2024).

Topical medications: Including glaucoma drops or preservative toxicity from eye drops containing preservatives (Andole & Senthil, 2023).

Skin diseases: On or around the eyelids, such as rosacea or eczema (Sobolewska, Schaller, & Zierhut, 2022).

Meibomian gland dysfunction: A common comorbidity with thickening and erythema of the eyelids and inadequate or altered secretions of meibomian glands (Bilgic et al., 2023).

Ophthalmic surgery: Refractive surgery, Cataract surgery, Keratoplasty, Lid surgery (Sabti Zechevikj & Raizada, 2022)

Chemical or thermal burns: Those that scar the conjunctiva (Napoli et al., 2019)

Ocular allergies and decreased androgen levels: Occurring with menopause and other conditions.

Computer or device usage: This may lead to decreased blinking when looking at the screen.

Excess or insufficient dosages of vitamins: Particularly vitamin A deficiency; can lead to xerophthalmia and the appearance of Bitot spots on the conjunctiva in severe cases.

Decreased sensation in the cornea: Due to: Long-term contact lens wear, Herpes virus infections.

Other causes of a neurotrophic cornea (Altinbas, Elibol, Fıratlı, Ayhan & Celebi, 2023)
Graft-versus-host disease, Autoimmune systemic diseases: Sjogren syndrome, Graves' ophthalmopathy.

Connective tissue disorders: Rheumatoid arthritis, Lupus, Thyroid disease (Choudhry et al., 2022)
Systemic conditions: Diabetes, Xerophthalmia, Hereditary disease, Gut dysbiosis, Multiple sclerosis and Nerve damage pathologies (García-Marqués Talens-Estarelles, C., García-Lázar, Wolffsohn & Cerviño, 2022)

Environmental factors: Including exposure to irritants such as: Chemical fumes, Cigarette smoke, Strong winds, High temperature, Pollution, High altitude and Low humidity (Tandon et al., 2020)
Behavioral habits: Smoking, Alcohol, Poor sleep, Unhealthy diet, Dislipidemia (Choi, Talens-Estarelles, García-Lázaro, S., Wolffsohn & Cerviño, 2020).

II. MATERIALS

This study utilised clinical, laboratory and data analysis materials required for subjects ocular examination, dry eye assessment, genetic analysis and statistical evaluation.

Clinical Examination Equipment

i. Self-illuminated Snellen’s visual acuity chart (by Alpha Optics) for distance visual acuity measurement.

ii. Snellen's near chart (by Alpha Optics) for near visual acuity measurement.
iii. Pen torch (by Oculite Pen) for external eye examination.
iv. Slitlamp biomicroscope (Huvitz Slitlamp 7000) for more detailed external eye examination.
v. Ophthalmoscope (Heine Direct Ophthalmoscope) for internal eye examination.

Dry Eye Assessment Materials

i. Fluorescein strip (by Nomax, Inc.) for staining the eyes before viewing with Slitlamp biomicroscpe while performing tear break up time test.
ii. Tetracaine hydrochloride 0.5% (by Fidson Pharmaceuticals) for numbing the eyes to prevent excessive eye tearing from irritation by the schirmer/paper strips.
iii. Schirmer strip with dimensions of 35 mm and 5 mm (by Nomax, Inc.) for identifying individuals with dry eyes.

Genetic Testing Materials

i. Buccal swab kits (by Copan Diagnostics) for collection of epithelial cells from the inner cheek for DNA extraction in a non-invasive manner.
ii. DNA extraction kit (QIAGEN QIAamp DNA Mini kit) to isolate and purify genomic DNA from buccal swab or blood samples for genetic analysis.
iii. Nano-drop Spectrophotometer (by Thermo Fisher Scientific) for DNA quantification for measuring the concentration and purity of extracted DNA to ensure it is suitable for downstream applications such as PCR and sequencing.
iv. PCR reagents (DNA polymerase, buffer and nuclease-free water) used to amplify specific DNA regions during polymerase chain reaction (PCR).
v. PCR thermal cycler (Bio-Rad T100) used to control temperature cycles required for DNA denaturation, primer annealing and DNA extension during PCR.
vi. Primers (short synthetic DNA sequences that bind to specific target DNA regions and initiate DNA amplification during PCR).
vii. Sequencing reagents- are chemicals required for DNA sequencing reactions including fluorescent nucleotides, enzymes and reaction buffers.

Clinical and laboratory consumables

i. Gloves (by soft care) to protect both participant and researchers from contamination and maintain aseptic conditions.
ii. Face masks (by soft care): to reduce the risk of contamination and ensure bio safety during specimen collection and laboratory procedures.
iii. Sterile swabs: used for collecting buccal cell samples or cleaning laboratory surfaces while preventing contamination.
iv. Pipettes used to accurately measure and transfer liquids during laboratory procedures.
v. Micropipette tips are disposable tips attached to micropipettes to transfer very small volumes accurately and prevent cross contamination.
vi. Microcentrifuge tubes for holding small volumes of samples during DNA extraction, PCR preparation and storage.
vii. Centrifuge tubes for sample preparation, centrifugation and storage of larger sample volumes.
viii. Distilled water used in reagent preparation, dilution of samples and cleaning laboratory equipment.
ix. Laboratory markers for labelling sample tubes, slides and containers for accurate identification and traceability throughout the study.

Validation of Instruments

The clinical instruments for this study were standard instruments approved by the Optometrist and Dispensing Opticians Registration Board of Nigeria (ODORBN). The laboratory instruments were standard instruments approved by Medical Laboratory Science Council of Nigeria (MLSCN) and validated by the department of Optometry and department of Medical Laboratory Science respectively.

Area of Study

The area of study for this research was Owerri Municipal, Imo State. Owerri is the capital city of Imo State in Nigeria, set in the heart of Igboland. Its Latitude and longitude coordinates are: 5.476310, 7.025853. It is also the state's largest city. It has an estimated population of 406,575 with Owerri Municipal population approximately 127,213 as at the last official population census since 2006 and is

approximately 551.00 square kilometres (212.743 sq mi) in area. Christianity is the dominant religion in Owerri Municipal. Owerri Municipal has a tropical wet climate. Rain falls for most months of the year with a brief dry season, this makes Owerri Municipal people great farmers. It is the chief trade centre for yams, cassava, corn, and palm products for a region of modified rainforest that also yields rubber for export.

Research Design

This design was purely clinical and laboratory based. The clinical aspect involved the recruitment and examination of participants who met the diagnostic criteria for dry eye disease. The laboratory component involved the collection and analysis of biological samples for genetic testing.

Population of Study

The study population comprised individuals diagnosed with dry eye disease who were resident in Owerri Municipal, Imo State, Nigeria. Participants were recruited from sponsored vision screening programs, medical outreaches and selected eye care facilities within the municipality. The target population included male and female patients aged 18 years and above who presented with symptoms and clinical signs of dry eye disease. Owerri municipal has a population of approximately 127,213 people according to the 2006 National Population Commission (NPC) census.

Inclusion Criteria

i. Adults aged 18-65 years.
ii. Individuals clinically diagnosed with dry eye disease by an eye care professional.
iii. Individuals who were residents of Owerri Municipal during the study period.
iv. Individuals who were willing to participate in the study.
v. Individuals willing to permit the collection of tear film samples from their eyes.

Exclusion Criteria

i. Subjects with history of ocular trauma or injuries that may alter ocular surface integrity.
ii. Subjects with history of eyelid abnormalities like ectropion and entropion.
iii. Subjects with history of Arthritis.
i. All pregnant women due to hormonal changes during pregnancy which may influence tear film physiology.
ii. Individuals unwilling to provide informed consent or complete all study procedures.

Sample Size Determination

The sample size for this study was determined using the Cochran formula for estimating a single population proportion:

$n = Z2pq/d2$

n = minimum required sample size
Z = standard normal deviate at 95% confidence level (1.96)
p = estimated prevalence of dry eye disease (50% or 0.50)
$q = 1 - p = 0.50$
d = margin of error (5% or 0.05)

Substituting the values into the formula:

$N=((1.96)2(0.50)(0.50))/((0.05)2)$
N= 120

Due to financial constraints, laboratory costs and number of eligible subjects available during the study period, all eligible and consenting subjects resulting in a sample size of 120 were enrolled.

Sampling Technique

Convenient sampling technique was used for this study. Population was selected by availability and declaration of interest; interested individuals participated after a due consent.

Procedure for Data Collection

Proper demographic data of subjects was taken including gender and age. Case history of patient was taken.

Visual acuity of subjects was taken at far and near. External examination was performed using pen torch and slitlamp biomicroscope. Internal examination was performed to rule out pathologies using the direct Ophthalmoscope.

The subjects were informed of the procedure. One drop of local anaesthetic eyedrop was instilled in both eyes to prevent excessive eye tearing from irritation by the paper strips.

The patient was asked to sit comfortably in the designated examination area. The clinician's hands were washed before conducting the test to ensure cleanliness. The Schirmer strip was removed from the package and folded around 5mm from the rounded inner end. The strip's folded end was then gently positioned in the lower fornix at the intersection of the middle and outer thirds of each eye's lower eyelid.

Subjects were instructed to close their eyes gently for 5 minutes. The timer was started once strip was in place and eyes shut. Schirmer strip was gently removed once the 5-minute period is over. Afterward, the moistened section's length is measured from the baseline mark to the region's endpoint. Less than 10mm is considered dry eye.

Tear Break up Time (TBUT) was measured by instilling Fluorescein 2% into the lower fornix of subject's eyes and examined under a slitlamp biomicroscope with a broad beam illumination and cobalt blue filter. The patient was asked to blink once and to keep their eyes open. The presence of black spots indicated dry spots on the tear film. TBUT is the time interval between the last blink and appearance of the first randomly-distributed dry spot. Dry spots in less than 10seconds is considered dry eye.

Subjects confirmed to have dry eye disease were sent to the laboratory for tear sample collection and genetic testing. Genetic variants responsible for dry eye disease (DNAJB6, MAML3, LINCO2267, MUC16, SIRPB3P, GAS2L3, KLF4, PAX6) and ocular surface inflammation (DNAJB6, MAML3, LINCO2267, MUC16) were identified and grouped accordingly.

The approach for the identification of the genetic variants underlying dry eye disease and ocular surface inflammation includes DNA and RNA sampling from subjects and subsequent DNA isolation from the samples utilizing commercial reagent kits.

Procedure for Data Analysis

Frequencies and descriptive tables were generated using the Statistical Package for Social Sciences (SPSS). The Pearson Chi-square and the student t-test statistics was used to explore relationships and associations for nominal and quantitative variables as appropriate. AP < .05 was considered statistically significant.

Ethical Consent

The approval of this research was obtained from the School of Health Technology, Federal University of Technology, Owerri. Participation in the study was entirely voluntary and participants were informed of their right to decline participation or withdraw from the study at any given stage. Confidentiality and anonymity were maintained throughout the study by assigning identification codes instead of recording participant's names. All information obtained during the study was treated as confidential and used solely for research purposes.

III. RESULTS

Gender Distribution of Subjects

Below shows the gender distribution of subjects used in the research. A total number of 120 Dry eye disease subjects participated in the research. There were 58 (48.3%) males and 62 (51.7%) females clinically diagnosed with Dry eye disease.

Table Showing Gender Distribution of Subjects

	Frequency (n)	Percentage (%)
Male	58	48.3
Female	62	51.7
Total	120	100

n=number of subjects, %=percentage of subjects

Age Distribution of Dry Eye Disease Subjects

Table below shows the age distribution of subjects that were used in the research. Subjects between 50-65 (36.7%) years of age were higher in number than other subjects. The least number of subjects fall within 18-29 (5%).

© AUG 2026 | IRE Journals | Volume 10 Issue 2 | ISSN: 2456-8880
DOI: https://doi.org/10.64388/IREV10I2-1722223

Table Showing Age Distribution of Dry Eye Disease Subjects

Age group	Frequency (n)	Percentage (%)
18-29	6	5
30-39	36	30
40-49	34	28.3
50-65	44	36.7
Total	120	100

n=number of subjects, %=percentage of subjects

Clinical Diagnosis of Dry Eye Disease (DED Clinical) and Ocular Surface Inflammation (OSI Clinical)

Table below shows clinical diagnosis of Dry eye disease and Ocular surface inflammation among subjects. A total number of 120 (100%) subjects were diagnosed with Dry eye disease, 72 (60%) were clinically diagnosed with ocular surface inflammation and 48 (40%) were clinically diagnosed of no ocular surface inflammation.

Table Showing Clinical Diagnosis of Dry Eye Disease (DED Clinical) and Ocular Surface Inflammation (OSI Clinical)

Condition	Present n (%)	Absent n (%)
Dry eye disease (clinical)	120 (100)	0 (0)
Ocular surface inflammation (clinical)	72 (60)	48 (40)

n=number of subjects, %=percentage of subjects

Genetic Diagnosis of Dry Eye Disease (DED Genetic) and Ocular Surface Inflammation (OSI Genetic)

Table below shows genetic diagnosis of Dry eye disease and Ocular surface inflammation among subjects. A total number of 120 (100%) subjects were diagnosed with Dry eye disease, 72 (60%) were genetically diagnosed with ocular surface inflammation and 48 (40%) were clinically diagnosed of no ocular surface inflammation.

Showing Genetic Diagnosis of Dry Eye Disease (DED Genetic) and Ocular Surface Inflammation (OSI Genetic)

Condition	Present n (%)	Absent n (%)
Dry eye disease (genetic)	120 (100)	0 (0)
Ocular surface inflammation (genetic)	72 (60)	48 (40)

n=number of subjects, %=percentage of subjects

Comparison between DED (Clinical) and DED (Genetic)

Table below shows the comparison between DED (clinical) and DED (genetic). All 120 (0%) subjects clinically diagnosed with Dry eye disease were also genetically diagnosed with Dry eye disease.

Showing Comparison between DED (Clinical) and DED (Genetic)

Condition	Present n (%)	Absent n (%)
Dry eye disease (clinical)	120 (100)	0 (0)
Dry eye disease (genetic)	120 (100)	0 (0)

n=number of subjects, %=percentage of subjects

Comparison between OSI (Clinical) and OSI (Genetic)

Table below shows the comparison between OSI (clinical) and OSI (genetic). All 72 (60) subjects clinically diagnosed with Ocular surface inflammation were also genetically diagnosed with Ocular surface inflammation while 48 subjects clinically diagnosed with no Ocular surface inflammation were genetically diagnosed with no Ocular surface inflammation.

Table showing Comparison between OSI (Clinical) and OSI (Genetic)

Condition (Genetic)	Present n (%)	Absent n (%)
Ocular surface inflammation (clinical)	72 (60)	48 (40)
Ocular surface inflammation (genetic)	72 (60)	48 (40)

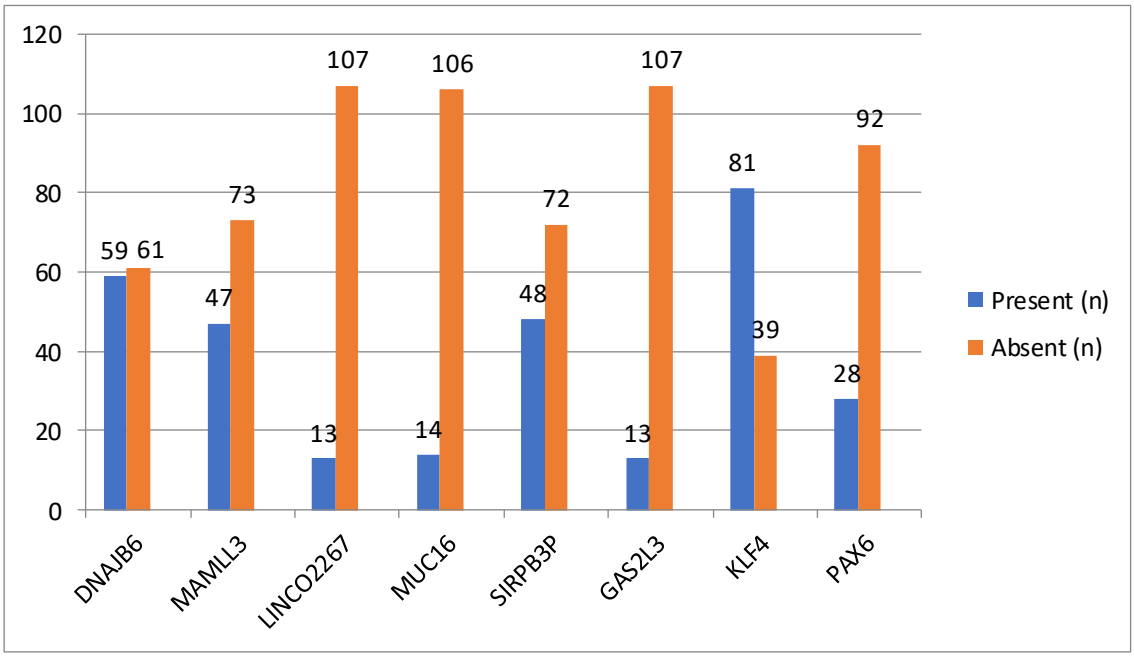
n=number of subjects, %=percentage of subjects

© AUG 2026 | IRE Journals | Volume 10 Issue 2 | ISSN: 2456-8880
DOI: https://doi.org/10.64388/IREV10I2-1722223

Frequency of Detected Genetic Variants

Figure below shows the frequency of detected genetic variants among DED subjects. KLF4 genetic variant was present in 81 (67.5%) subjects and absent in 39 (32.5%) subjects making it the highest genetic variant found in DED subjects. LINCCO2267 and GAS2L3 genetic variants were equally present in 13 (10.8%) subjects and absent in 107 (89.2%) subjects making them the least genetic variant present among the DED subjects.

Figure showing Frequency of Detected Genetic Variants



IV. TESTING OF HYPOTHESES

Testing the First Hypothesis

HO1: There is no association between genetic variants and dry eye disease.

SPSS data analysis result showing p-value for testing the first hypothesis

Dry Eye Disease	Pearson's Correlation	P-value
Clinical Diagnosis – Genetic Variants	1.00	0.00

From SPSS version 23 data output, data analysis using the Pearson Product Moment Correlation Coefficient at 0.05 level of significance revealed a p-value of 0.00. Since p(0.00) <0.05, the null hypothesis is rejected and the alternate accepted.

HA1: There is an association between genetic variants and dry eye disease.

Testing the Second Hypothesis

HO2: There is no association between genetic variants and ocular surface inflammation.

SPSS data analysis result showing p-value for testing the second hypothesis

Ocular Surface Inflammation	Pearson's Correlation	P-value
Clinical Diagnosis – Genetic Variants	1.00	0.00

From SPSS version 23 data output, data analysis using the Pearson Product Moment Correlation Coefficient at 0.05 level of significance revealed a p-value of 0.00. Since p(0.00) < 0.05, the null hypothesis is rejected and the alternate accepted.

HA2: There is an association between genetic variants and ocular surface inflammation.

V. DISCUSSION

The prevalence of dry eye disease and ocular surface inflammation observed in this study underscores the substantial burden of ocular surface disorders within the study population. The high proportion of subjects exhibiting overlapping DED and OSI (60%) reinforces the concept that dry eye disease is not merely a tear deficiency disorder but an inflammatory condition affecting the ocular surface. These findings are consistent with epidemiological studies by Craig et al. (2017), who reported that inflammatory mechanisms underlie both aqueous-deficient and evaporative forms of dry eye. Similarly, Schaumberg et al. (2003), demonstrated that a significant proportion of dry eye patients exhibit subclinical or overt inflammatory changes.

However, some population-based studies conducted in Asian cohorts by Hsu et al. (2024) have reported lower overlap rates between DED and OSI. Hsu et al. (2024) also reported that this discrepancy may be attributable to differences in diagnostic criteria, environmental exposure, ethnic genetic background, and access to healthcare. They further stated that the use of combined clinical and molecular assessment may have increased sensitivity for detecting inflammatory involvement.

The enrichment of variants in DNAJB6, MAML3, LINC02267, MUC16, SIRPB3P, GAS2L3, KLF4, and PAX6 among DED subjects provides strong evidence for a genetic contribution to the disease susceptibility.

LINCO2267 and GAS2L3
LINCO2267 acts as a competing endogenous RNA (ceRNA). It regulates inflammation in DED through the NONMMUT047964.2/miR-671-5p/Egr-1 axis while GAS2L3 play a role in maintaining the integrity of the ocular surface epithelium and its response to stress.

It is observed in this research that LINCO2267 and GAS2L3 are the least detected among genetic variants of DED (being present in 13 DED subjects and absent in 107 DED subjects) while LINCO2267 is found to be the least detected among the genetic variants responsible for OSI in DED subjects (also detected in 13 and absent in 107 DED subjects) .

MUC16
MUC16 encodes a membrane-associated mucin that contributes to the epithelial glycocalyx and tear film stability. Disruption of this barrier predisposes the ocular surface to desiccation and inflammation.
It is observed in this research that MUC16 is the second to the least in frequency (being present in 14 DED subjects and absent in 106 DED subjects). This finding aligns with studies by Argüeso and Gipson (2011) and Mantelli and Argüeso (2008), which identified mucin dysregulation as a central mechanism in dry eye pathology.

KLF4 and PAX6
KLF4 and PAX6 are both key transcription factors in corneal epithelial differentiation; this further supports their role of impaired epithelial renewal in DED according to Collinson et al., (2003). This research agrees with Collinson et al., (2003) that reduced KLF4 expression is the cause of compromised epithelial barrier function in DED. This work is equally in agreement with Swamynathan et al., (2007) which states that PAX6 mutations are associated with corneal surface abnormalities. This is evident as the presence of PAX6 is low in the genetic variants detected among DED subjects.

DNAJB6
DNAJB6 functions as a molecular chaperone involved in cellular stress responses. Its association with OSI supports findings from in vitro ocular surface stress models, where chaperone proteins are upregulated in response to hyperosmolar and inflammatory stress. Its association with DED and OSI is evident in the frequency of detected genetic variants of DED and OSI. It is observed in this research that DNAJB6 is the most detected among the genetic variants responsible for OSI.

MAML3
MAML3 is involved in Notch signalling, a pathway regulating epithelial–immune interactions. Its

association with OSI aligns with emerging evidence linking Notch dysregulation to chronic inflammatory disorders. Its association with DED and OSI is evident in the frequency of detected genetic variants of DED and OSI. This is in line with the research by Stevenson et al., (2011) where he described Dry eye disease as an immune-mediated ocular surface disorder.

The significant overrepresentation of DNAJB6, MAML3, LINC02267, and MUC16 variants among OSI subjects highlights shared molecular pathways between dry eye disease and ocular surface inflammation.