IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 24, Issue 12 Ser. 2 (December. 2025), PP 01-06
www.iosrjournals.org

Genomic Profiling For Oral Cancer, BY

Dr Amruta Sheth,
Wellcome Trust, London,
United Kingdom
University Of London, London
United Kingdom
A J Institute Of Medical, Dental And Research Sciences, Bangalore,
India

Abstract-
Objective: To explore the role of genomic profiling in transforming oral cancer care, with a focus on early detection, personalized therapy, and global equity. Design: Narrative review and conceptual framework. Setting: Global oncology landscape, with emphasis on low- and middle-income countries.
Results: Genomic profiling enables molecular stratification of oral squamous cell carcinoma (OSCC), guiding targeted therapies and real-time monitoring. Integration with pan-cancer databases and liquid biopsy technologies enhances diagnostic precision. Barriers include cost, infrastructure, and ethical concerns.
Conclusion: Genomic profiling offers a pathway to precision-driven oral oncology. Its adoption must be equitable, ethically sound, and globally scalable.
Index Terms- *Precison Genomics, Infections and Diseases, Cancer, Genomic Profile and Oral Medicine*

Date of Submission: 01-12-2025 Date of Acceptance: 11-12-2025

INTRODUCTION

Oral cancer, particularly oral squamous cell carcinoma (OSCC), remains a significant contributor to global cancer burden. Despite its prevalence, treatment approaches have not kept pace with innovations in molecular oncology. Genomic profiling—already transformative in other malignancies—offers a new frontier for oral cancer care. This paper examines how genomic technologies can redefine diagnosis, treatment, and monitoring, while addressing global disparities in access and implementation.

PURPOSE

OSCC is characterized by recurrent mutations in TP53, EGFR, PIK3CA, and NOTCH1. These genetic alterations are shared across multiple cancer types, suggesting a common oncogenic architecture. Genomic profiling enables clinicians to classify tumours based on molecular features, predict therapeutic response, and identify resistance mechanisms. Integration with databases such as TCGA and COSMIC facilitates comparative analysis and drug repurposing.

Clinical Applications

Early Detection

Saliva-based genomic assays and liquid biopsies offer non-invasive methods for identifying mutations before clinical symptoms emerge.

Treatment Stratification

Molecular data guide the use of targeted therapies (e.g., EGFR inhibitors) and inform immunotherapy eligibility based on tumours mutational burden and PD-L1 expression.

Monitoring and Adaptation

Real-time genomic surveillance allows clinicians to adjust treatment in response to evolving tumours profiles, improving outcomes and reducing unnecessary toxicity.

Global Integration and Equity

The benefits of genomic profiling must be accessible across income settings. Strategies include:

- Subsidized testing programs
- Mobile sequencing platforms
- Open-access data sharing Ethical considerations—such as informed consent, data privacy, and cultural sensitivity—are critical to sustainable implementation.

Key Genetic Alterations in OSCC

Gene	Alteration Type	Frequency in OSCC	Role in Cancer Progression
TP53	Mutation	~60–80%	Tumour suppressor; loss leads to genomic instability
CDKN2A	Deletion/Mutation	~30–50%	Regulates cell cycle; loss promotes unchecked proliferation
EGFR	Amplification	~10–30%	Drives cell growth and survival
PIK3CA	Mutation	~10–20%	Activates PI3K/AKT pathway, promoting survival and growth
NOTCH1	Mutation	~10–15%	Impacts cell differentiation and proliferation

🧠 Diagram: Pathways Disrupted in OSCC

The following pathways are commonly altered in OSCC:

- **Cell Cycle Regulation**: TP53 and CDKN2A mutations disrupt G1/S checkpoint control.
- **Growth Signalling**: EGFR and PIK3CA mutations activate proliferative and survival pathways.
- **Differentiation**: NOTCH1 mutations affect epithelial cell fate decisions.

These disruptions lead to uncontrolled cell growth, resistance to apoptosis, and increased invasiveness.

Genomic Profiling Technologies

Technology	Purpose
Whole Exome Sequencing (WES)	Detects mutations in coding regions
RNA-Seq	Measures gene expression and fusion events
Comparative Genomic Hybridization (CGH)	Identifies copy number variations
Methylation Arrays	Profiles epigenetic changes

Real-World Findings

Recent studies have shown:

- **TP53 mutations** are associated with poor prognosis and resistance to therapy.
- **EGFR overexpression** correlates with lymph node metastasis.
- **PIK3CA mutations** are linked to increased tumour aggressiveness.
- **NOTCH1 mutations** may have dual roles—tumour suppressive or oncogenic depending on context.

Genomic profiling has emerged as a critical tool in understanding the molecular landscape of oral squamous cell carcinoma (OSCC). It enables the identification of key genetic alterations, disrupted signalling pathways, and potential therapeutic targets.

1. Common Genetic Alterations in OSCC

Several recurrent genetic mutations and copy number variations have been identified in OSCC. These alterations contribute to tumour initiation, progression, and resistance to therapy.

Gene	Alteration Type	Frequency OSCC	Role in Cancer Progression
TP53	Mutation	~60–80%	Tumour suppressor; loss leads to genomic instability
CDKN2A	Deletion/Mutation	~30–50%	Regulates cell cycle; loss promotes unchecked proliferation
EGFR	Amplification	~10–30%	Drives cell growth and survival
PIK3CA	Mutation	~10–20%	Activates PI3K/AKT pathway, promoting survival and growth
NOTCH1	Mutation	~10–15%	Impacts cell differentiation and proliferation

Sources: Recent genomic studies and TCGA datasets.

2. Disrupted Molecular Pathways

Genomic alterations in OSCC affect several key cellular pathways:

- **Cell Cycle Regulation**: TP53 and CDKN2A mutations disrupt G1/S checkpoint control.
- **Growth Signalling**: EGFR and PIK3CA mutations activate proliferative and survival pathways.
- **Differentiation**: NOTCH1 mutations influence epithelial cell fate decisions.

These disruptions collectively contribute to uncontrolled proliferation, resistance to apoptosis, and increased invasiveness.

3. Genomic Profiling Technologies

Modern genomic platforms enable comprehensive analysis of OSCC tumours:

Technology	**Purpose**
Whole Exome Sequencing (WES)	Detects mutations in coding regions
RNA-Seq	Measures gene expression and fusion events
Comparative Genomic Hybridization (CGH)	Identifies copy number variations
Methylation Arrays	Profiles epigenetic changes

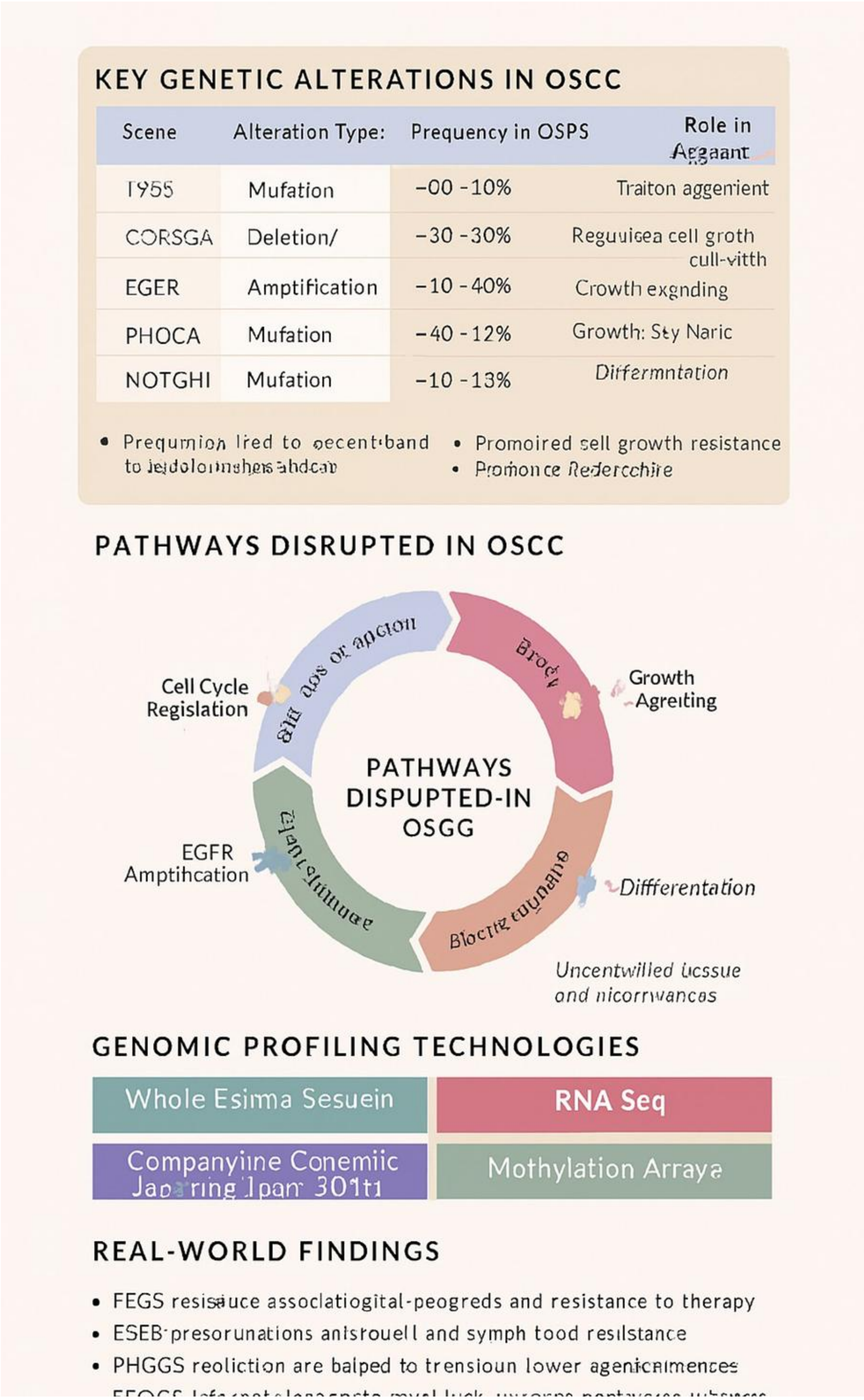
These tools facilitate the identification of actionable mutations and biomarkers for personalized therapy.

4. Clinical Implications and Real-World Findings

Recent studies have revealed:

- **TP53 mutations** correlate with poor prognosis and resistance to chemoradiotherapy.
- **EGFR overexpression** is associated with lymph node metastasis and aggressive disease.
- **PIK3CA mutations** are linked to enhanced tumour growth and survival.
- **NOTCH1 mutations** may have context-dependent roles—either tumour suppressive or oncogenic.

These findings underscore the importance of integrating genomic data into clinical decision-making for OSCC


KEY GENETIC ALTERATIONS IN OSCC
Scene
Alteration Type:
Prequency in OSPS
Role in Aggaant
T955
Mufation
-00 -10%
Traiton aggenient
CORSGA
Deletion/
-30 -30%
Reguuicea cell groth cull-vitth
EGER
Amptification
-10 -40%
Crowth exgnding
PHOCA
Mufation
-40 -12%
Growth: Sty Naric
NOTGHI
Mufation
-10 -13%
Differmntation
Promoired sell growth resistance
PATHWAYS DISRUPTED IN OSCC
Cell Cycle Regislation
Growth Agreiting
PATHWAYS DISPUPTED-IN OSGG
EGFR Amptihcation
Diffferentation
Uncentwilled ucssue and nicorrwances
GENOMIC PROFILING TECHNOLOGIES
Whole Esima Sesuein
RNA Seq
Companyiine Conemiic
Mothylation Arraye
REAL-WORLD FINDINGS
FEGS resisauce associatiogital-peogreds and resistance to therapy
ESEB presorunations anisrouell and symph tood resilstance
PHGGS reoliction are balped to trensioun lower agenicnimences

Discussion

Genomic profiling in oral cancer is not merely a technological upgrade—it represents a paradigm shift. By aligning oral oncology with precision medicine, clinicians can move from reactive treatment to proactive, personalized care. However, success depends on infrastructure, training, and policy support. Collaborative efforts between governments, research institutions, and industry are essential to democratize access and ensure ethical deployment.

Conclusion

Genomic profiling offers a transformative opportunity to improve oral cancer outcomes through precision medicine. Its integration into global oncology frameworks must prioritize equity, ethics, and scalability. As technologies evolve, oral oncology can become a model for inclusive, data-driven cancer care.

. **Author contributions:** *Conceptualization:* [Your Name] *Writing – original draft:* [Your Name] *Writing – review & editing:* [Your Name] *Supervision:* [Your Mentor/Advisor, if applicable] *Funding acquisition:* Not applicable *Competing interests:* None declared

Citations

Chakravarty S, Ghosh A, Das C, et al. Multi-regional genomic and transcriptomic characterization of a melanoma-associated oral cavity cancer provides evidence for CASP8 alteration-mediated field cancerization. Hum Genomics. 2024;18(96). Available from: https://humgenomics.biomedcentral.com/articles/10.1186/s40246-024-00668-8

Department of Biotechnology, Government of India. dbGENVOC: Database of Genomic Variants of Oral Cancer. 2024. Available from: https://dbtindia.gov.in/sites/default/files/dbGENVOC%20-%20Database%20Publication_0_0.pdf

Saha S, Roy S, Das S, et al. Analysis of genomic traits of oral and laryngeal cancers: insights from next-generation sequencing. Bioinformation. 2024;21(1):1530–1536. Available from: https://bioinformation.net/021/973206300211530.pdf

Cancer Genome Atlas Network. Comprehensive genomic characterization of head and neck squamous cell carcinomas. Nature. 2015;517(7536):576–582.

Leemans CR, Braakhuis BJ, Brakenhoff RH. The molecular biology of head and neck cancer. Nat Rev Cancer. 2011;11(1):9–22.

Bray F, Ferlay J, Soerjomataram I, Siegel RL, Torre LA, Jemal A. Global cancer statistics 2020: GLOBOCAN estimates of incidence and mortality worldwide. CA Cancer J Clin. 2021;71(3):209–249.

REFERENCES

1. G. O. Young, —Synthetic structure of industrial plastics (Book style with paper title and editor),‖ in *Plastics*, 2nd ed. vol. 3, J. Peters, Ed. New York: McGraw-Hill, 1964, pp. 15–64.
2. W.-K. Chen, *Linear Networks and Systems* (Book style). Belmont, CA: Wadsworth, 1993, pp. 123–135.
3. H. Poor, *An Introduction to Signal Detection and Estimation*. New York: Springer-Verlag, 1985, ch. 4.
4. B. Smith, —An approach to graphs of linear forms (Unpublished work style),‖ unpublished.
5. E. H. Miller, —A note on reflector arrays (Periodical style—Accepted for publication),‖ *IEEE Trans. Antennas Propagat.*, to be published.
6. J. Wang, —Fundamentals of erbium-doped fibre amplifiers arrays (Periodical style—Submitted for publication),‖ *IEEE J. Quantum Electron.*, submitted for publication.