Letter to Editor

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Reflections on the Efficacy of QF-PCR for Prenatal Screening in the Iranian Population: Implications for Global Practices

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Dear Editor,

I am writing to share my reflections on the recent article by Mazaheri et al. regarding the effectiveness of Quantitative Fluorescent Polymerase Chain Reaction (QF-PCR) for prenatal screening in the Iranian population. The study reveals several compelling insights that warrant further discussion within the medical community.[1]

First and foremost, the high concordance rate of 99.8% between QF-PCR and conventional karyotyping is a notable achievement. This finding indicates that QF-PCR may be a reliable alternative for detecting common aneuploidies such as trisomy 21 and 18. The rapid turnaround time associated with QF-PCR could significantly reduce the logistics burden of traditional karyotyping, thus allowing for quicker clinical decision-making and potentially improving outcomes for expecting parents.

The analysis of heterozygosity among the 25 Short Tandem Repeats (STR) markers is another important facet of this research. Pinpointing informative markers specifically for the Iranian population can enhance the sensitivity of prenatal screening tests, emphasizing the critical need for tailored approaches to genetic screening based on genetic diversity. Such population-specific data is essential as it underscores the importance of localized research in effective genetic screening practices.

The study provides valuable insights into the relationship between nuchal translucency (NT) and trisomy risk. The correlation

between increased fetal NT and higher detection rates of trisomy 21 reinforces the necessity of routine NT measurements as part of prenatal assessments. It is especially crucial for high-risk groups, such as mothers of advanced age, and supports the integration of NT measurements with genetic testing to optimize prenatal care. However, the study also highlights discrepancies between QF-PCR and karyotyping, particularly in cases of sex chromosomal rearrangements and mosaicism. These findings raise critical questions about the diagnostic capacity of QF-PCR for specific conditions that may not present clearly in standard karyotyping. Consequently, while QF-PCR demonstrates efficacy as a primary screening tool, it underscores the importance of confirmatory testing through conventional methods in certain instances.

The implications of this research contribute significantly to the understanding of prenatal screening in non-Western populations, an area often underrepresented in existing literature. By focusing on the Iranian population, this study enhances our understanding of the broader applicability of genetic screening practices across diverse genetic backgrounds.

The study presents significant advancements in detecting aneuploidies within the Iranian population by evaluating the effectiveness of QF-PCR compared to conventional karyotyping. It focuses on the heterozygosity of 25 short tandem repeat (STR) markers, identifying the most informative ones for prenatal screening. It also reports an impressive concordance rate of 99.8% between QF-PCR and karyotyping. This high level of agreement suggests a promising potential for QF-PCR as a stand-alone method in prenatal diagnostics. Among its advantages, QF-PCR is noted for its rapid, low-cost, and convenient nature, significantly reducing workload and turnaround time for results. The comprehensive analysis enhances clinical decision-making by improving the accuracy of detecting common aneuploidies. However, the study acknowledges limitations, including a sample size that may not fully represent the Iranian population, potentially affecting the generalizability of the findings. Additionally, there are discrepancies between QF-PCR and karyotyping results, particularly concerning sex chromosomal rearrangements and cases of mosaicism, indicating that QF-PCR may overlook specific chromosomal abnormalities. The research also does not address the potential effects of maternal cell contamination on karyotyping results, which could further influence the accuracy of the conclusions drawn.

In conclusion, I endorse the study's advocacy for integrating QF-PCR as a standard practice in prenatal testing, especially in resource-limited settings. The insights gained can lead to improved prenatal diagnostic protocols that are both accurate and culturally relevant. Future research exploring the long-term impacts of utilizing QF-PCR in diverse populations could yield even more significant advancements in global prenatal care.

Conflict of Interest

The authors declare no conflicts of interest.

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Ethical Considerations

None.

Author's Contribution

Writing- original draft preparation, H.N.; editing the manuscript, S.A.D. Both authors have read and agreed to the published version of the manuscript.

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Reference

1. Mazaheri M, Yavari M, Behravesh S, NikKhah H, Ekraminasab S. Prenatal Screening Using QF-PCR and Karyotyping with an Evaluation of Short Tandem Repeats Markers in the Iranian Population. World J Peri Neonatol 2023; 6(1): 1-9.