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LIFE SCIENCE FOR A SUSTAINABLE FUTURE

(Seminar Proceedings of the International Conference on Current Trends in life Science for a Sustainable Future)

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Molecular Analysis of *IRF6* Gene In Non-syndromic Cleft Lip And Palate: A Pilot Study

Reema Rose Alappat[1,2], Sunish K.S[1], PV Narayanan[3], P.R. Varghese[2], Alex George[2*]

The most common orofacial birth abnormality, Non-syndromic cleft lip with or without cleft palate (NSCLP), has a wide range of occurrence across the world, which is generally related to ethnic and environmental variances. NSCLP susceptibility is ethnicity-dependent, and the genetic basis of susceptibility to NSCLP is expected to differ amongst groups. The earlier studies show that *IRF6* assumes a focal part in NSCLP This study investigated the contribution of *IRF6* gene polymorphisms in NSCLP formation. Following inclusion and exclusion criteria, 20 NSCLP patients and 20 controls were recruited. DNA isolation and genotyping of the single nucleotide polymorphisms in exons 3 and 4 of the *IRF6* gene in two groups were determined by PCR and Sanger sequencing. In the present study, we identified an SNP rs7552506 in Intron 3 of the *IRF6* gene. Allele frequencies of rs7552506 show that the control sample and the patient sample showed 32% and 32% G allele respectively. However, none of the models with different genotype combinations of these polymorphisms, such as codominant, dominant, recessive, or over-dominant, exhibited any connection with NSCLP.

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This is to certify that ~~Dr./Mr./~~Ms. Reema Rose Alappat, Research Scholar, Maharajas College, Trivandrum has participated/presented (Oral/Poster) in the "International Conference on *Current Trends in Life Science for a Sustainable Future*" organized by Mar Ivanios College (Autonomous), Thiruvananthapuram from 4th - 6th August, 2022.

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Mutation Analysis of *WNT4* Gene In 46, XX Disorders of Sexual Development

Ragitha T.S.[1,2], Sunish K.S.[1], Sareena Gilvaz[3], Saley Daniel[3], P.R. Varghese[2], Suresh KumarR.[2]*

Disorders of Sexual Developments (DSDs) are rare congenital diseases characterised by aberrant development of the internal and external genitalia. DSDs are divided into three groups based on their chromosomal components: 46,XX DSD, 46,XY DSD, and Sex Chromosomal DSD. The absence of Sex determining region of the Y (*SRY*) gene and the action of gene like Wingless-type MMTV integration site family, member 4 (*WNT4*) leads to female reproductive structure development. Genetic variations of this gene results in 46,XX DSD. The molecular basis of 46,XX DSD still remains unclear, underscoring the current need for research. In the present study we analysed the role of *WNT4* gene mutations in 46,XX DSD patients. In our study we recruited 100 adolescent girls with primary amenorrhea (aged 14 to 22 years). Based on the cytogenetic and *SRY* gene analysis, 32 cases were included for *WNT4* mutation analysis. PCR sequencing was performed for all coding exons of *WNT4*. Bioinformatic tools like Mutation Taster, Human Splicing Finder and microRNA Data Base (miRDB) were used. We observed single nucleotide variations in three patients. One had a known synonymous polymorphism (rs544988174). miRDB data

revealed the absence of microRNA regulatory site in this region. Other two carried a nucleotide substitution in intronic regions and it does not affect the normal splicing mechanism. In the future, further investigation on these variants will be required in more samples.

Key Words: Disorders of Sexual Development, 46,XX DSD, *WNT4*, *SRY*, Primary Amenorrhea.

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