The Identification of Genetics Factor of Non-Syndromic Orofacial Cleft in the Southeast Asian Population

Intended use

As a part of Virtual Presentation in

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This Presentation is based on

a study conducted by A.T. Yusuf, G. Nuranti$_3$, Z.A. Al-

Outline

1. Introduction
2. Background
3. Research Question
4. Methodology
5. Result and Findings
6. Conclusions
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“

Research is to see what everybody else has seen, and to think what nobody else has thought.

Albert Szent-Gyorgyi

Introduction

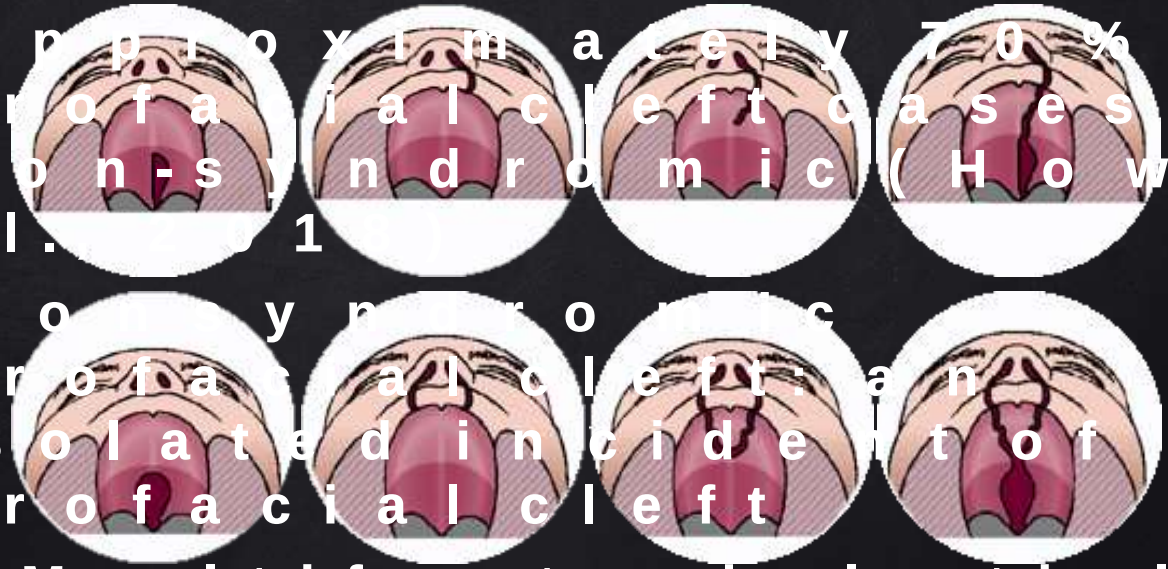
W h a t t h e s t u d y i s a b o u t

What is nonsyndromic orofacial cleft?

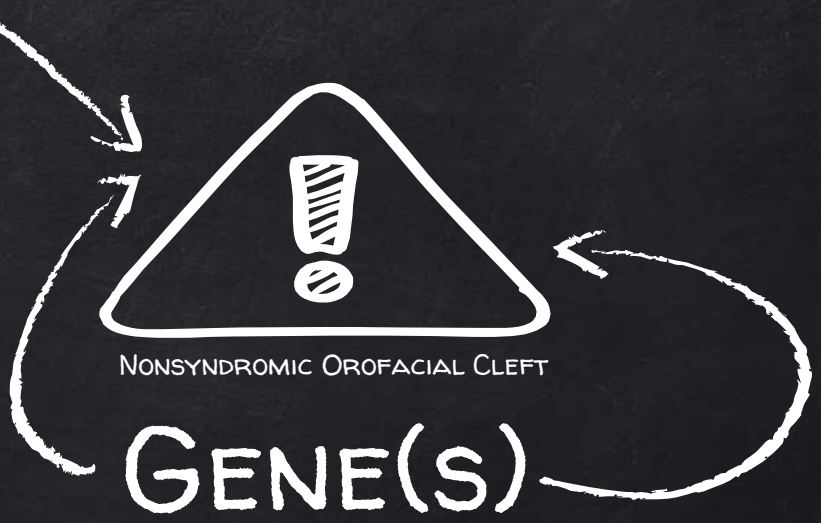
- Orofacial cleft (OFC):
  - the most common craniofacial birth defects in humans (Jugessur et al., 2009)
  - A major congenital structure anomaly (Schutte & Murray, 1999):
    - significant lifelong morbidity

What is nonsyndromic orofacial cleft?

- Approximately 70% of orofacial cleft cases are non-syndromic (Howe et al., 2021)
- Nonsyndromic orofacial cleft: an isolated incident of orofacial cleft
- Multifactorial etiology (Mehrotra et al., 2022)



Environmental Factors



Multifactorial etiology

(Adeyemo et al., 2012; Dixon et al., 2011), 2

What is nonsyndromic orofacial cleft?

- Approximately 70% of orofacial cleft cases are non-syndromic (Howe et al., 2018)
- Nonsyndromic orofacial cleft: an isolated incident of orofacial cleft
  - Multifactorial etiolo
  - Different prevalence throughout differen

Prevalence

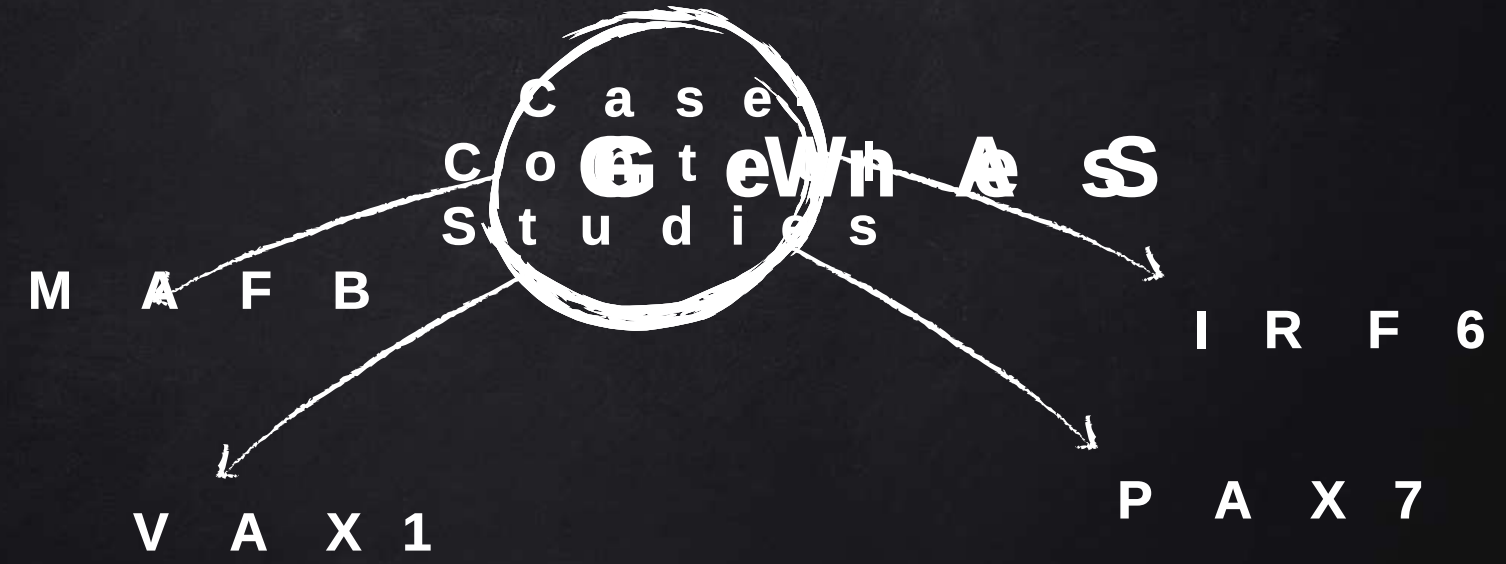
Globally
1.2 per 1,000 live birth

(Murray, 2010; Liu et al., 2016; Vieira et al., 2019; Mutua et al., 2018)

Background

Why the study was conducted?

Nonsyndromic Orofacial Cleft Research



(Birnbaum et al., 2009; Graf et al.,

Nonsyndromic Orofacial Cleft Research

Gene(s)? → Gene Mapping → European / American

(Marazita & Leslie, 2013)

Research Questions

What is the objective of the study?

Research Question

Southeast Asian Orofacial Cleft Gene(s)?

Main Gene Candidate(s)?

Methodology

How the study was conducted

Methodology

Identification

Screening & Eligibility Test

Meta-Analysis

n t i f i c a t i o n

K e y w o r d s

M e S H T e r m s

A l l F i e l d s

1 9 8 0 - 2 0 1 7

Screening

Tittle & Abstract

Duplicate Remove

Further Screening

Identified Result

Inclusion Criteria :

Case Report

Case Control

1980-2017

Cross Sectional Studies

Significant P value

Cohort Studies

Exclusion Crite

Abstract & Title

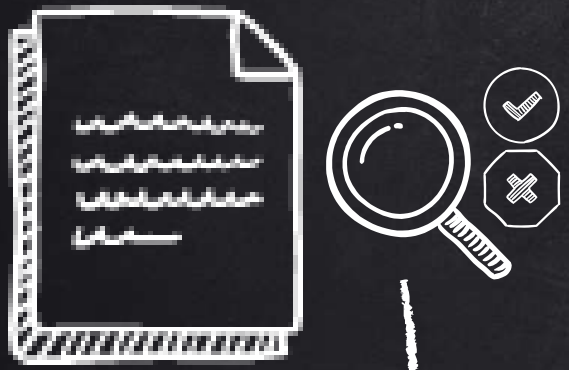
Study with no C

Environment R

Animal Study

Insufficient Dat

Eligibility Test



Inclusion Criteria

Exclusion Criteria

Scale Value → 0 - 9

Study quality :

The Newcastle-Ottawa Scale (NOS)

1. Selection
2. Comparability
3. Exposure

(Sabbagh et al., 2015)

Low risk of bias/ 'good quality

Modest risk of bias/ 'fair quali

Substantial risk of bias/ 'poor qu

Result & Findings

W h a t t h e s t u d y f o u n d

Identification

Screening

Records identified through database searching (PUBMED) (n = 158)

Records identified through other sources (SCHOLAR) (n = 715)

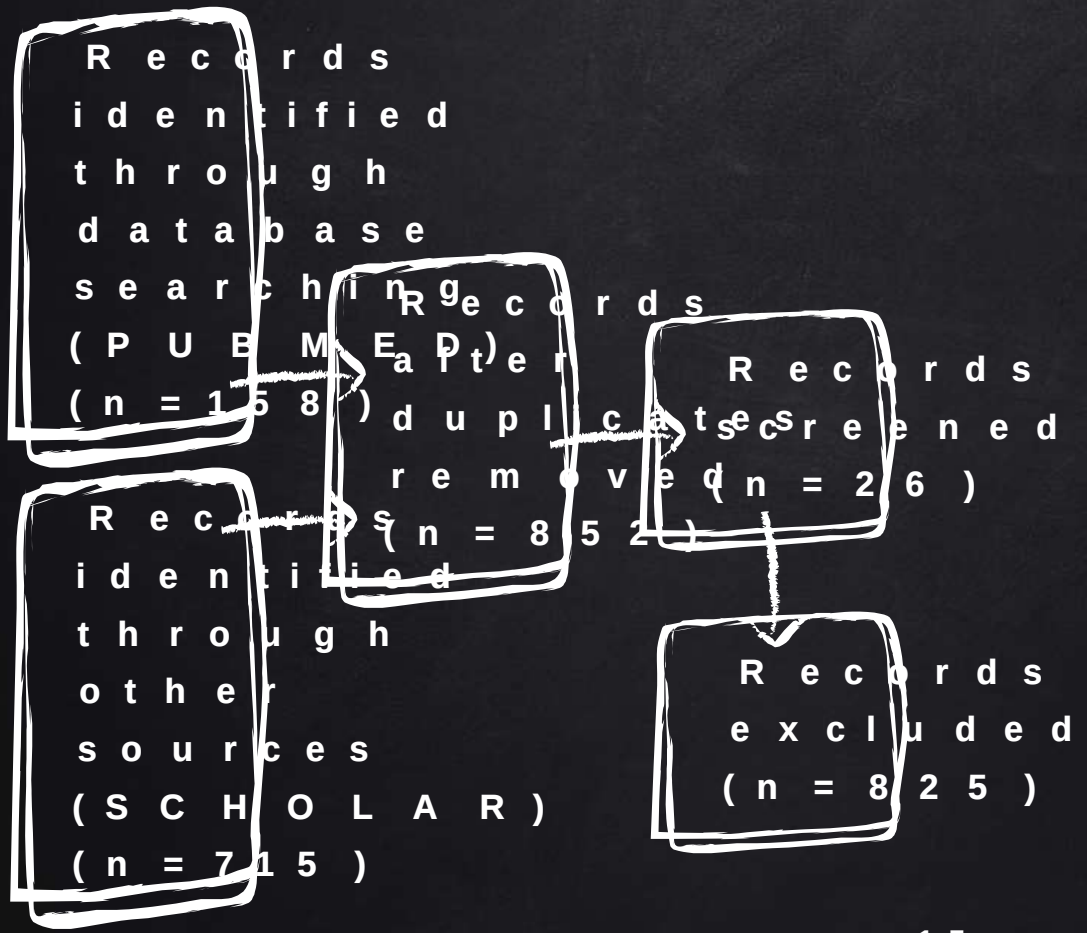
Records after duplicates removed (n = 852)

Records screened (n = 26)

Records excluded (n = 825)

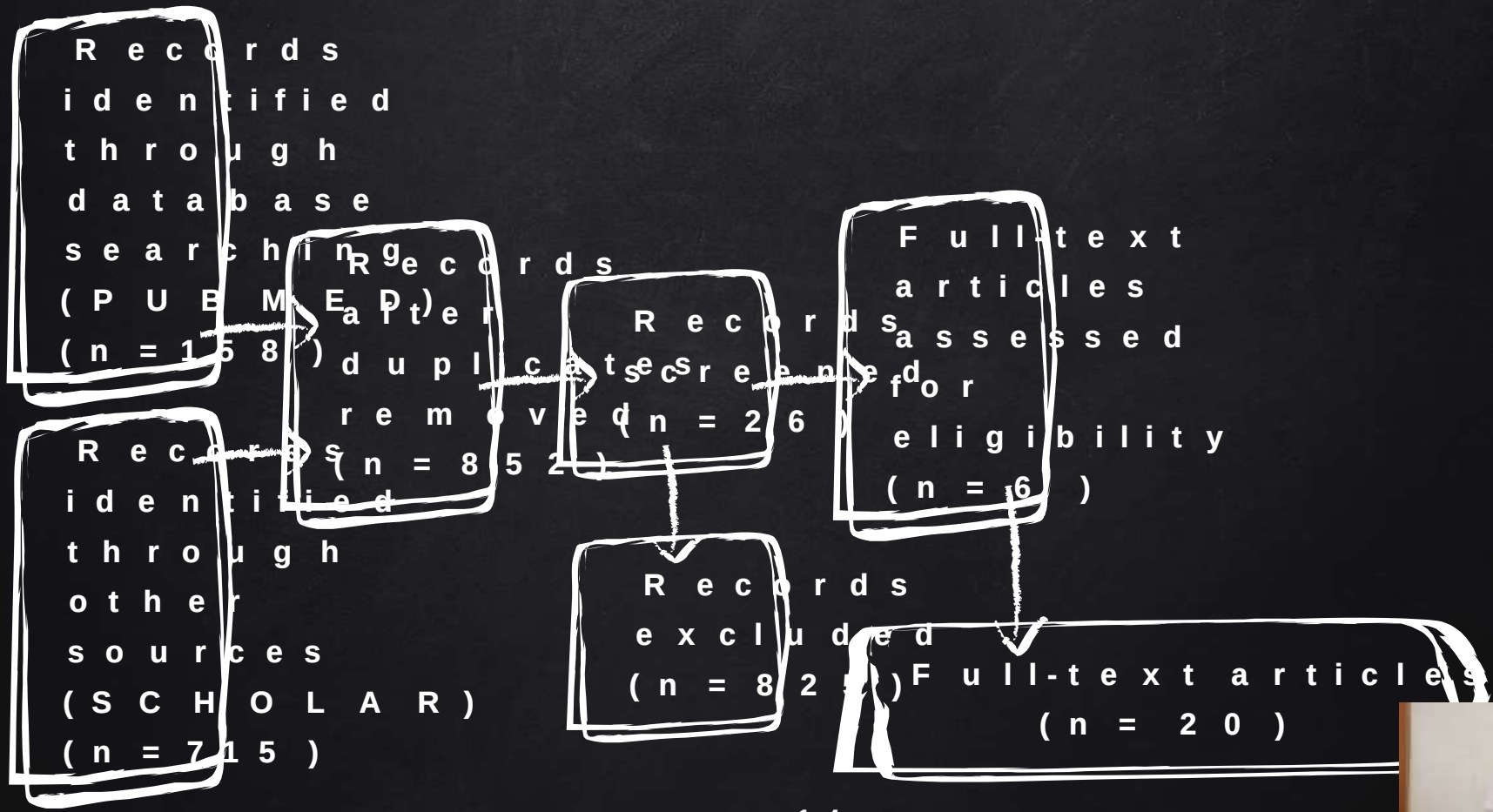
Exclusion criteria and number of excluded studies from The First Selection

Identification Screening Eligibility

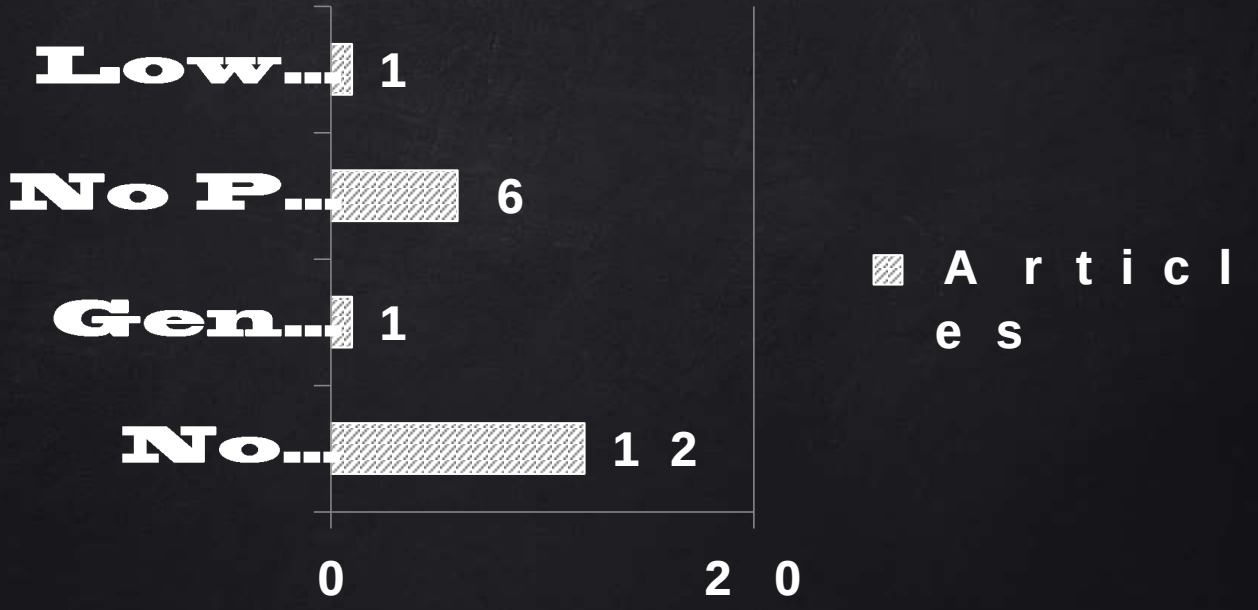


Genes				
M T H F R	T G F A	B M P 4	M M P 1 3	E G F
F G F R 2	I R F 6	M S X 1	T B X 2 2	P V R L 1
P A X 7	A B C A 4	E V C 2	C R I S P L D 2	6 p 2 4
M A F B	8 q 2 4	C O L 8 A 1 / F I L I P 1 L	N T N 1	V A X 1
N O G	S L C 2 A 9	F O X $F_{1\ 7}2$	C D H 1	L P N 2

Identification Screening Eligibility



Exclusion Criteria



Identification | Screening | Eligibility | Incl

Records identified through database searching (PUBMED) (n = 158)

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Records after duplicates removed (n = 852)

Records screened (n = 26)

Records excluded (n = 826)

Full-text articles assessed for eligibility (n = 6)

Full-text articles excluded (n = 20)

Studies included in qualitative synthesis (n = 6)

Studies included in quantitative synthesis (meta-analysis) (n = 6)

Included studies after the second screening stage

Author	Year	Gene	Total NOS Score	P Value
Schultz et al.	2004	TGFA	8	0.01
Srchomthong et al.	2005	IRF6	9	0.02
Tongkobpetch et al.	2006	MSX1	8	0.05
Salahshourifar et al.[14]	2011	MSX1	8	0.0

MSX1 (MUSCLE SEGMENT HOMEOBOX 1) GENE

- Plays a crucial role in epithelial-mesenchymal tissue interactions
- Regulation gene in the cellular proliferation, differentiation and cell death (Bendall & Abate-Shen, 2000)
- Mutation contribute in nonsyndromic orofacial cleft (Jugessur et al.,

Conclusions

W h a t c a n b e c o n c l u d e d

Conclusions

Orofacial Cleft Genes in Southeast Asia

TGFA, IRF6, MSX1, CDH1 and ARHGAP29

Main Gene Candidate

MSX1

Acknowledgement

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Bibliography

Dixon MJ, Marazita ML, Beaty TH, Murray JC. Cleft lip and palate: understanding genetic and environmental influences. Nat Rev Genet 2011;12(3):167-78.

Shaw GM, Croen LA, Curry CJ. Isolated oral cleft malformations: associations with mater and infant characteristic

Thank You!

Any questions?

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