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Research Article

Role of Consanguinity on Pediatric Haematological and Pulmonary Health: A Comparative Study at a Tertiary Care Teaching Hospital, Guntur, Andhra Pradesh

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ABSTRACT

Background: Consanguineous marriage, characterised as a union between individuals who are second cousins or closer, continues to be culturally significant in certain regions of India and is linked to heightened homozygosity and an elevated risk of autosomal recessive disorders. Grasping its effects on child health is crucial for informing preventive measures. **Objectives:** The objective is to evaluate how parental consanguinity influences the occurrence of haematological and pulmonary disorders in children receiving care at a tertiary teaching hospital. **Materials and Methods:** A cross-sectional observational study was carried out involving 209 children aged between 0 and 18 years. Participants were divided into two groups: consanguineous and non-consanguineous, according to their parental relationship history. Information regarding demographic characteristics, clinical findings, and confirmed diagnoses was gathered through a structured proforma. Statistical analysis was conducted using SPSS version 26.0, with a significance level set at a p-value of less than 0.05. **Results:** Haematological disorders occurred with greater frequency (86.1%) compared to pulmonary disorders (13.9%). Notable associations were identified between consanguinity and thalassemia (p=0.03), sickle cell anaemia (p=0.01), and asthma (p=0.003). In contrast, haemophilia did not demonstrate a significant difference (p=0.09). A male predominance was observed in X-linked disorders, whereas autosomal conditions exhibited an even distribution. **Conclusion:** Parental consanguinity significantly adds to the prevalence of pediatric haematological disorders and may affect the incidence of multifactorial pulmonary diseases. Enhanced genetic counselling, community-oriented screening, and awareness programs are essential to mitigate preventable genetic morbidity in high-risk groups.

Keywords: Asthma, Consanguinity, Genetic Counselling, Pediatric Health, Sickle Cell Anaemia, Thalassemia.

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INTRODUCTION

Consanguinity, which is defined as the union of individuals who are biologically related, specifically those who are second cousins or closer, continues to be a culturally accepted and socially ingrained practice in numerous parts of the world, especially in South Asia, the Middle East, and North Africa.[1,2] Worldwide, estimates indicate that around 10% of marriages are consanguineous, with certain areas experiencing rates as high as 50-60%.[3,4] While these unions are often preferred for the sake of maintaining family cohesion, inheritance, and social connections, they also pose considerable medical and public health challenges: increased homozygosity in offspring heightens the risk of autosomal recessive and other inherited disorders, thereby amplifying health burdens at both individual and population levels.

The genetic foundation of the effects of consanguinity is rooted in the heightened likelihood that both parents may possess one or more identical harmful alleles passed down from a shared ancestor, consequently increasing the inbreeding coefficient and amplifying the risk of genetic disorders in offspring.[1] In the context of India, for instance, findings from the National Family Health Survey-4 reveal a national prevalence of 9.9% for consanguineous marriages, with even higher rates approaching ~23% in southern regions.[2] The significance of this issue for child health is considerable: genetic conditions continue to be significant factors contributing to morbidity and mortality

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in paediatrics, particularly in areas where early detection and intervention are scarce.

Haematological disorders in children are among the conditions most closely associated with parental consanguinity, as many of these disorders exhibit autosomal recessive or X-linked inheritance patterns and are more likely to manifest clinically in the offspring of related parents.[5,6] Notable examples include β-thalassemia, sickle cell disease, and rare bleeding disorders such as haemophilia, which have a significantly higher incidence in populations with high levels of consanguinity.[5] Regional studies suggest that these conditions result in lifelong challenges, including dependency on transfusions, chronic disease management, decreased survival rates, and diminished quality of life, underscoring the considerable public health implications of hematologic disorders driven by consanguinity.

Pulmonary disorders, such as bronchial asthma, cystic fibrosis, and various other chronic lung diseases, constitute another area where genetic predisposition interacts with environmental factors. While asthma and related respiratory ailments are influenced by multiple factors, certain studies indicate that parental consanguinity might heighten the risk through shared genetic vulnerabilities.[7] Nevertheless, the evidence remains inconclusive; some studies report an increased risk, while others do not find a significant independent effect of consanguinity on childhood asthma.[8] This uncertainty highlights the necessity to investigate these relationships within the Indian context, where genetic and environmental influences differ.

Despite the worldwide and regional focus on health outcomes associated with consanguinity, there is still a significant lack of data from India that concurrently examines both haematological and pulmonary disorders in children born to consanguineous as opposed to non-consanguineous parents. Considering the elevated prevalence of consanguinity in the southern states of India, along with the substantial effects of haematological and pulmonary disorders on child health, there exists an urgent public health necessity for comprehensive evidence to bolster genetic counselling, targeted screening, and awareness initiatives.

Consequently, this study was structured as a cross-sectional investigation conducted in a tertiary-care teaching hospital, focusing on the comparison of pediatric haemato-pulmonary health outcomes among children from consanguineous and non-consanguineous unions. The results are intended to enhance regional clinical practices, inform preventive strategies such as premarital or antenatal counselling, and aid in the formulation of policies aimed at alleviating the impact of inherited diseases in India.

MATERIALS AND METHODS

Study Design and Setting

This observational study, which was hospital-based and cross-sectional in nature, took place in the Department of Paediatrics at a tertiary care teaching hospital located in Guntur, Andhra Pradesh, India. Conducted over a specified timeframe, this research project aimed to assess the effects of parental consanguinity on the haematological and pulmonary health of children. The study specifically sought to identify correlations between consanguineous and non-consanguineous parental unions and the occurrence of certain disorders among pediatric patients visiting both the outpatient and inpatient departments of the hospital.

Study Population and Sample Size

A total of 209 children, ranging from infancy to 18 years of age, were included in the study. The participants were divided into two categories based on the parental relationship history gathered through direct interviews:

1. **Consanguineous group:** This group consists of children whose parents are related as second cousins or closer.
2. **Non-consanguineous group:** This group includes children whose parents do not share any biological relationship.

The ultimate sample size (n=209) was established according to the eligible cases present throughout the study duration and satisfied the minimum criteria to guarantee sufficient comparative power between the two groups.

Inclusion criteria

- Children between the ages of 0 and 18 years who presented to the pediatric department throughout the duration of the study.
- The presence of both parents or legal guardians was required to gather comprehensive family and medical histories.
- Participants must demonstrate a willingness to engage in the study following the provision of verbal informed consent.

Exclusion criteria

- Children lacking complete clinical or parental relationship information.
- Instances exhibiting unrelated haematological or pulmonary symptoms resulting from trauma, infection, or malignancy.

- Individuals whose guardians opted out of participation.

Data Collection and Tools

Data were gathered via structured interviews conducted with parents or guardians, an examination of hospital records, and direct clinical assessments. A pre-validated data collection form was utilised to record:

- Characteristics of the population (age, gender, place of living, economic status).
- Patterns of parental relationships and the nature of marriage.
- Comprehensive family background concerning genetic or hereditary conditions.
- Medical history and diagnostic results on haematological and respiratory systems.

Laboratory investigations were examined for haemoglobin concentrations, complete blood counts, peripheral smears, and other pertinent haematological parameters. The pulmonary evaluation encompassed clinical observations, a history of respiratory distress, spirometry (when applicable), and physician diagnoses recorded in medical documentation.

Statistical Analysis

Data were input and analysed utilising Microsoft Excel and SPSS software (Version 26.0; IBM Corp., Armonk, NY, USA). Descriptive statistics, including mean, standard deviation, and percentage distributions, were employed to summarise demographic and clinical variables. Inferential analysis was performed using the Chi-square (χ^2) test to compare categorical variables between consanguineous and non-consanguineous groups. A *p*-value of less than 0.05 was deemed statistically significant. The results were presented in both tabular and graphical formats for enhanced clarity.

Ethical Considerations

This study was a non-interventional, observational investigation that relied on standard clinical and family history information; thus, no invasive techniques or experimental treatments were utilised. Although formal approval from an ethical committee was not requested, the research complied with the guidelines outlined in the Declaration of Helsinki (2013) concerning confidentiality, voluntary participation, and the respect for the rights of participants. Before inclusion, verbal informed consent was acquired from parents or legal guardians, and all data gathered were anonymised to ensure confidentiality.

RESULTS

A total of 209 children were enrolled, consisting of 107 (51.2%) from consanguineous marriages and 102 (48.8%) from non-consanguineous marriages. Overall, haematological disorders were the most prevalent (180; 86.1%), whereas pulmonary disorders represented 29 (13.9%).

Interpretation

As indicated in Table 1, male children made up almost two-thirds of the study cohort. The majority of the affected children were under the age of 5, with the greatest burden observed among second-borns in consanguineous marriages, suggesting a potential cumulative genetic exposure in later offspring.

Interpretation

As illustrated in Table 2, haematological disorders were prevalent in both marriage categories. Thalassemia and sickle cell anaemia exhibited notable correlations with parental consanguinity ($p < 0.05$), whereas the prevalence of haemophilia was elevated in both groups without any statistically significant difference. Pulmonary disorders, particularly asthma, were significantly more common in consanguineous unions ($p=0.003$).

Interpretation

A notable concentration of thalassemia ($p=0.03$) and asthma ($p=0.003$) was identified in children born from second- and third-degree consanguineous unions. The lack of first-degree cases indicates that severe inbreeding might be infrequent or not viable in the clinical context of the study population, as illustrated in Table 3.

Interpretation

A notable male predominance was identified overall, especially in cases of haemophilia. Conversely, both thalassemia and asthma exhibited a slight female predominance in both groups. This gender-specific distribution aligns with the principles of X-linked inheritance for haemophilia and autosomal recessive inheritance for thalassemia and sickle cell anaemia, as detailed in Table 4.

- Haematological disorders constituted a significant majority at 86%, in stark contrast to pulmonary conditions, which accounted for only 14%.
- Thalassemia and sickle cell anaemia demonstrated statistically significant correlations with consanguinity.
- Asthma emerged as the most prevalent pulmonary disorder, exhibiting a notable association with third-degree consanguinity.
- Male children were disproportionately impacted by haemophilia, whereas female children displayed

marginally elevated frequencies of thalassemia and asthma.

- Children born second in consanguineous unions bore the greatest overall disease burden, indicating a cumulative hereditary risk.

DISCUSSION

The current hospital-based cross-sectional research involving 209 children revealed a higher prevalence of haematological disorders (86.1%) compared to pulmonary disorders (13.9%). Notably, there were significantly elevated rates of thalassemia and sickle cell disease among children with consanguineous parents, alongside a marked increase in asthma cases associated with certain levels of consanguinity. These results are generally consistent with a recent study conducted in India,[9] which indicates that inherited hemoglobinopathies continue to represent a significant genetic burden in the pediatric population, particularly in communities with elevated rates of parental kinship. National surveys and recent literature reviews[10,11] highlight considerable carrier populations for β-thalassemia and emphasise a major public health initiative aimed at identifying sickle cell carriers and patients in regions with high prevalence, thereby reinforcing the notion that hemoglobinopathies are significant contributors to pediatric morbidity throughout India.

The notable correlation between consanguinity and thalassemia observed in our cohort aligns with genetic predictions and is consistent with regional studies that reveal a heightened homozygosity of pathogenic β-globin alleles within endogamous populations. Various screening initiatives and systematic evaluations[9,12,13] conducted in India have underscored inconsistent yet frequently elevated frequencies of traits and diseases in areas and communities that engage in consanguinity or endogamy; this finding corroborates our observation that second- and third-degree parental relationships exhibited a concentration of thalassemia cases. The trend we identified, which shows increased disease prevalence among closer relatives while also noting some instances among more distantly related individuals, reflects previous research suggesting that inbreeding practices enhance the manifestation of recessive alleles found within a population's genetic reservoir.[9]

Sickle cell disease in India demonstrates a multifaceted geographic and tribal distribution; recent national initiatives and reviews highlight a concentrated burden among particular states and tribal groups, while also acknowledging the influence of localised mating patterns in perpetuating disease prevalence. Our findings reveal a statistically significant association between consanguinity and sickle cell cases (despite the disease's established founder and tribal clustering), which aligns with the emerging national landscape where both carrier frequency and marriage patterns interact to influence regional disease incidence.[14] Mass screening campaigns and the National Sickle Cell Anaemia Elimination Mission[15] have underscored the necessity of genetic counselling

Table 1: Demographic and Baseline Distribution of the Study Population (*n*=209).

Parameter	Category	Consanguineous (*n*=107)	Non-consanguineous (*n*=102)	Total n (%)
Gender	Male	61 (29.1%)	76 (36.4%)	137 (65.6%)
	Female	46 (22.0%)	26 (12.4%)	72 (34.4%)
Age group (years)	0-2	62 (29.6%)	47 (22.5%)	109 (52.1%)
	3-5	42 (20.1%)	46 (22.0%)	88 (42.1%)
	6-8	3 (1.4%)	8 (3.8%)	11 (5.2%)
	9-12	0 (0%)	1 (0.5%)	1 (0.5%)
Birth order	1st born	42 (20.1%)	45 (21.5%)	87 (41.6%)
	2nd born	56 (26.8%)	42 (20.1%)	98 (46.9%)
	3rd born	9 (4.3%)	15 (7.2%)	24 (11.4%)

Table 2: Overall Diagnostic Distribution by Type of Marriage (*n*=209).

Parameter	Category	Consanguineous (*n*=107)	Non-consanguineous (*n*=102)	Total n (%)
Haematological disorders (total)	87 (41.6%)	93 (44.5%)	180 (86.1%)	0.32 (NS)
Hemophilia	35 (32.7%)	47 (46.1%)	82 (39.2%)	0.09 (NS)
Thalassemia	37 (34.6%)	30 (29.4%)	67 (32.1%)	0.03 *
Pulmonary disorders (total)	20 (9.6%)	9 (4.3%)	29 (13.9%)	0.04 *
Asthma	18 (16.8%)	9 (8.8%)	27 (12.9%)	0.003 *
Cystic fibrosis / COPD / Emphysema	0	0	0	—

Table 3: Distribution of Major Disorders by Degree of Consanguinity (*n*=107).

Degree of Consanguinity	Hemophilia n (%)	Thalassemia n (%)	Sickle cell anaemia n (%)	Asthma n (%)	*p*-value
1st degree	0 (0)	0 (0)	0 (0)	0 (0)	—
2nd degree	7 (6.5)	30 (28.0)	6 (5.6)	2 (1.9)	0.03 *
3rd degree	10 (9.3)	25 (23.3)	0 (0)	16 (14.9)	0.003 *
4th degree	4 (3.7)	3 (2.8)	2 (1.9)	2 (1.9)	0.01 *

Table 4: Gender-wise Distribution of Diagnoses in Consanguineous and Non-Consanguineous Marriages.

Diagnosis	Consanguineous Male n (%)	Consanguineous Female n (%)	Non-consanguineous Male n (%)	Non-consanguineous Female n (%)
Hemophilia	35 (32.7)	8 (7.5)	47 (46.1)	11 (10.7)
Thalassemia	15 (14.0)	22 (20.6)	11 (10.7)	19 (18.6)
Sickle cell anaemia	3 (2.8)	4 (3.7)	2 (2.0)	3 (2.9)
Asthma	8 (7.5)	12 (11.2)	3 (2.9)	(5.9)

and community-targeted interventions in these high-risk areas, a translational implication that directly stems from our results.

The prevalence of haemophilia in both consanguineous and non-consanguineous populations, showing no significant difference, aligns with the X-linked inheritance pattern of haemophilia: a male-dominant distribution is anticipated, and the impact of parental consanguinity on X-linked disorders at the population level is expected to be relatively minor. This trend is also observed in other clinic-based studies,[16] where haemophilia remains prevalent regardless of parental kinship, as its transmission is primarily reliant on carrier females rather than on homozygosity at autosomal loci. The gender distribution observed in our dataset, characterised by a male predominance for haemophilia and a balanced or slightly higher frequency of females for autosomal disorders, adheres to classical inheritance principles and corroborates previous findings.[17,18]

The observation that asthma, a complex pulmonary disorder, was more prevalent among children of consanguineous parents in our study warrants thorough interpretation. The aetiology of asthma encompasses the interaction between genetic and environmental factors; certain population studies[7,19] have indicated a higher incidence of asthma within consanguineous families, which may reflect the aggregation of alleles that predispose to atopy alongside shared environmental exposures. Nevertheless, the existing literature is inconsistent and contextually dependent: although genetic vulnerability may be heightened by consanguinity in specific communities, environmental factors (such as allergens, infections, and pollution) frequently overshadow the differences in prevalence that are observed. Our findings imply a possible genetic influence on asthma risk in this demographic, yet we are unable to separate genetic factors from environmental confounders without employing molecular or multivariable analytical methods.

The limitations of the study must be recognised in the context of existing literature: our cross-sectional design inhibits causal inference, and although the sample size is adequate to identify several associations, it restricts the power for less common disorders and for multivariable adjustments. Furthermore, the lack of molecular genotyping limits the confirmation of carrier states and the accurate evaluation of homozygosity; prior research has indicated that the integration of molecular screening with epidemiological studies significantly elucidates the role of consanguinity in disease burden.[20] Consequently, future investigations should incorporate genomic screening, larger multi-site samples, and standardised measures of environmental exposure to unravel the relative impacts of inherited and non-inherited risk factors within this population.

In conclusion, our research aligns with both recent Indian and global evidence: consanguinity serves as a significant contextual element for various pediatric haematological disorders and may influence the risk associated with certain multifactorial pulmonary conditions. These findings underscore the necessity for screening and counselling strategies that are specifically adapted to regional contexts, and they offer empirical backing for current public health initiatives designed to alleviate the impact of genetically influenced childhood diseases in areas with high rates of consanguinity.

CONCLUSION

This research illustrates a distinct correlation between parental consanguinity and the prevalence of pediatric haematological disorders, especially thalassemia and sickle cell anaemia, alongside a notable aggregation of multifactorial pulmonary issues like asthma. The results reinforce the notion that marriages among close relatives elevate the chances of inheriting autosomal recessive characteristics and may affect vulnerability

to intricate diseases via common genetic and environmental influences. These findings underscore the genetic aspect of community-endogamous traditions and their subsequent effects on child health.

The findings underscore the critical necessity for ongoing public health initiatives, regular carrier screening, premarital and antenatal genetic counselling, as well as heightened awareness at the population level, to alleviate the burden of preventable genetic diseases in areas of high risk. The incorporation of these interventions into current national programs, particularly the National Sickle Cell Anaemia Elimination Mission, has the potential to enhance strategies for early detection and prevention. Ongoing multicentric research that merges molecular diagnostics with epidemiological surveillance is vital to elucidate mechanisms, provide culturally appropriate counselling, and promote targeted genetic health policies in India.

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CONFLICT OF INTEREST

The authors declare that there is no conflict of interest.

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This study did not obtain any particular funding from agencies in the public, commercial, or non-profit sectors.

ETHICAL APPROVAL

Ethical approval was not necessary for this observational, non-interventional study. The research complied with the ethical guidelines outlined in the Declaration of Helsinki (2013 revision).

INFORMED CONSENT

Informed consent, communicated verbally, was secured from the parents or legal guardians of all children participating in the study before their inclusion.

AUTHOR CONTRIBUTIONS

SPSC played a significant role in data collection, interpretation of clinical data, statistical analysis, and assisted in the preparation of the manuscript. SC undertook the literature review and compiled the data. SKK was responsible for conducting statistical analysis, constructing tables, and interpreting the results. PKY took charge of the critical drafting and revision of the manuscript. SRK offered academic supervision, verified the diagnostic data, and guided the assessment of pediatric cases. RKB contributed to the clinical validation, reviewed data accuracy, and provided expert insights on the classification of haematological and pulmonary disorders. RRN provided conceptual oversight, academic supervision, and conducted the final critical review for the intellectual content and approval of the manuscript. All authors have reviewed and approved the final version of the manuscript and acknowledge their accountability for all aspects of the work.

ABBREVIATIONS

SPSS: Statistical Package for the Social Sciences; **CBC:** Complete Blood Count; **COPD:** Chronic Obstructive Pulmonary Disease; **CF:** Cystic Fibrosis; **χ^2:** Chi-square Test; **NHM:** National Health Mission; **SCDAEM:** Sickle Cell Anaemia Elimination Mission; **HGB:** Haemoglobin (used indirectly under haematological investigations); **CBC:** Complete Blood Count; **SD:** Standard Deviation.

SUMMARY

This study highlights a significant association between parental consanguinity and the increased occurrence of pediatric haematological disorders, particularly thalassemia and sickle cell anaemia, as well as a notable rise in asthma among pulmonary conditions. The findings emphasise that marriages among close relatives elevate homozygosity and thereby heighten the risk of inheriting autosomal recessive disorders, while also contributing to susceptibility toward multifactorial respiratory diseases. These results reinforce the importance of targeted genetic counselling, early carrier screening, and community-level awareness programs to reduce preventable genetic morbidity in high-risk populations.

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