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CONSAGUINITY AND ITS ASSOCIATION WITH GENETIC DISORDERS: A DECADE OF RESEARCH IN PAKISTAN

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ABSTRACT

Consanguinity means marriages within blood relations, mostly spouses are first or second cousins. This type of marriage is found throughout human history. Consanguinity causes genetic disorders like neurogenetic disorders, heart, dental anomalies, blood, skin, muscular, metabolic, and structural disorders. Additionally, it is also noted that prenatal and postnatal negative outcomes are very high in consanguineous marriages as compared to other populations. The use of modern genetic sequencing techniques, comparative analysis of consanguinity in different regions, psychosocial and ethical implications, public health recommendations, and future research directions in Pakistan are also underscored in this review. The prevalence of consanguinity is higher in Pakistan as compared to other countries in

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the world due to cultural and religious backgrounds. These investigations uncover the sensitivity and future consequences of this practice. This review will highlight the prevalence of consanguinity and its association with genetic disorders in Pakistan. The last ten years, from 2015 to 2025, published research work on consanguinity and genetic disorders is included in this review.

Key Words: Consanguinity, Genetic disorders, Prevalence, Psychosocial, Sequencing, Pakistan.

Introduction

Consanguineous marriage, in which spouses have blood relations, is found throughout human history. But this type of marriage creates medical issues like genetic disorders due to reproduction of blood-related pairs; genetic mutations are passed on into the next generation (Khayat et al., 2024). It has been investigated that the risk of congenital disorders in the offspring is found to be very high when parents are blood-related due to cultural and religious perspectives. Pakistan is considered at the top for consanguineous marriages. So region of common interests for medical researchers, biologists, physicians, and social scientists who deal with congenital abnormalities inherited from consanguineous parents (Ahmad et al., 2023). Monogenic disease is a disease caused by defects in genes. 10000 diseases found in the world are monogenic, and globally, 10/1000 live births. It is estimated that 350 million people are affected by monogenic genetic disorders globally. Thalassemia, sickle cell anaemia, colour blindness, haemophilia, deafness, autism, etc., are monogenic hereditary disorders. Pakistan is included in the countries that are at high risk of monogenic disorders. 80 per cent consanguinity is found in Pakistan (Aslamkhan, 2015). Consanguinity is the major factor for congenital anomalies and autosomal recessive disorders. Consanguinity is a very common tradition in different global areas in the world. Consanguineous marriages have been present in early human culture in history. Consanguinity is a common cause of the prevalence of different genetic diseases in human communities. It is also an urgent issue in Pakistan. Inherited disorders of familiar molecular genetic origin are identified by their genotype and phenotype, but heterogeneity is also observed, which is dependent. Consanguinity rate is increasing, so autosomal recessive diseases express more than one human genetic disorder in leading to overlapping phenotypes. Next-generation sequencing has new insights into these issues. On the opposite side, the effect of consanguinity on malignancies and multifactorial disorders is not so distinct (Ijaz et al., 2022; Temaj et al., 2022). Most of the consanguinity observed age at time of marriage was between 13-22 and 34-32 years for females and males, respectively. Percentage of illiterate wives (43.8%), while their husbands (36.1%) have secondary level education, many wives (87.6%) are housewives, and 42.8% of husbands were earning, and 60.9% of spouses (60.9%) belong to middle-class social status (Ijaz et al., 2022; Fazal et al., 2023). Consanguineous marriage is considered to save property, to grow social links, and to grow kinship family structures. Consanguinity is linked with the risk of chromosomal disorders. Pakistan has with highest rates of global consanguinity, with about 60% of all marriages having blood relations. A report to study the effect of modernization on consanguinity in Kabirwala, Pakistan, indicates that consanguinity is still prevalent in

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Kabirwala, with 77% of consanguineous marriages (Irfan et al., 2023; Hayat et al., 2022). At Rahim Yar Khan (RYK) District is a mix of both local and migrated communities in Southern Punjab, Pakistan. Study showed that consanguineous unions (CU) were 58.46 %, and the inbreeding coefficient (IC-F) = 0.0355. The consanguinity was higher in people from rural areas, illiterate or having a religious/Madrasah education only, and having a nuclear family structure. The rate of CU is also higher in people whose male spouses perform unskilled manual or skilled manual work, and had cousin marriages in the parents' generation. Multivariate logistic regression analyses revealed that the first cousin unions (constituted 52 % of all marriages), parallel-cousin and patrilineal unions were in the majority (54 and 57 %, respectively), and father's brother's daughter type rate was 31 % (Riaz et al., 2016).Congenital anomalies (CA) or birth defects are more serious in developing countries. In a study at a tertiary care facility in Rawalpindi, Pakistan. In which 517 independent cases with CA were analyzed. Neurological disorders (39.1%), neuromuscular disorders (21.1%), limb defects (13.5%), musculoskeletal defects (7.4%), blood disorders (4.3%), orofacial defects (3.9%), metabolic disorders (3.7%), and Others (7.1%). The sporadic (72.5%) vs familial cases (27.5%). Among total cases, 70% cases were involved in parental consanguinity (Shaheen et al., 2023; Hayat et al., 2018). congenital anomalies (CA) in Baluchistan province of Pakistan was studied with a group of 1185 independent patients studied with CA males were 71%., neurological disorders with highest prevalence (n=317; 27%), limb disorders (n=161; 14%), blood-heart disorders (n=159; 13%), neuromuscular disorders (n=156; 13%), sensorineural/ear disorders (n=140; 12%), eye/visual disorders (n=90; 8%), musculoskeletal disorders (n=83; 7%), ectodermal disorders (n=31; 3%) .61% CA were sporadic 39% were familial; and parental consanguinity was diagnosed in 51% cases (M. Q. Khan et al., 2024; Imad et al., 2020). A cross-sectional study was conducted at District Head Quarter Hospital, Okara, aged 19 to 55 years, with and without consanguinity results showed a significant link between consanguinity and congenital anomalies at birth, time showed p p-value of 0.002(S. Khalid et al., 2019; Rafaqat et al., 2023). Public knowledge and attitudes toward genetic testing and inherited disorders in Pakistan were observed. 78.89% participants knew about genetic testing, while 87.15% showed willingness in genetic testing if a facility is provided. Also, 87.15% agreed with pre-marital genetic testing. Results showed a solid public interest in genetic services (Shareef et al., 2025; Shoaib et al., 2022). A genetic study was carried out in Buner, Charsadda, Mardan, Nowshera, Peshawar, and Swabi districts to observe the congenital anomalies. Study showed congenital anomalies like neurological disorders, limb defects, sensorineural defects, musculoskeletal defects, visual impairments, hemoglobinopathies, ectodermal disorders, cardiovascular anomalies, and orofacial anomalies. Most cases had a sporadic presentation (68%) and an isolated occurrence (72%), while 58% cases with parental consanguinity (Naqvi et al., 2024; Hayat et al., 2020). Socioeconomic and demographic factors behind consanguineous marriages showed that uneducated and poor family backgrounds are more associated with consanguinity, with a higher prevalence of miscarriages, abortions, and stillbirths in consanguinity as compared to non-consanguinity. The trend analysis showed that the trend of non-consanguineous marriages is increasing in Pakistan. Consanguinity was found (80% in 1980) while 65

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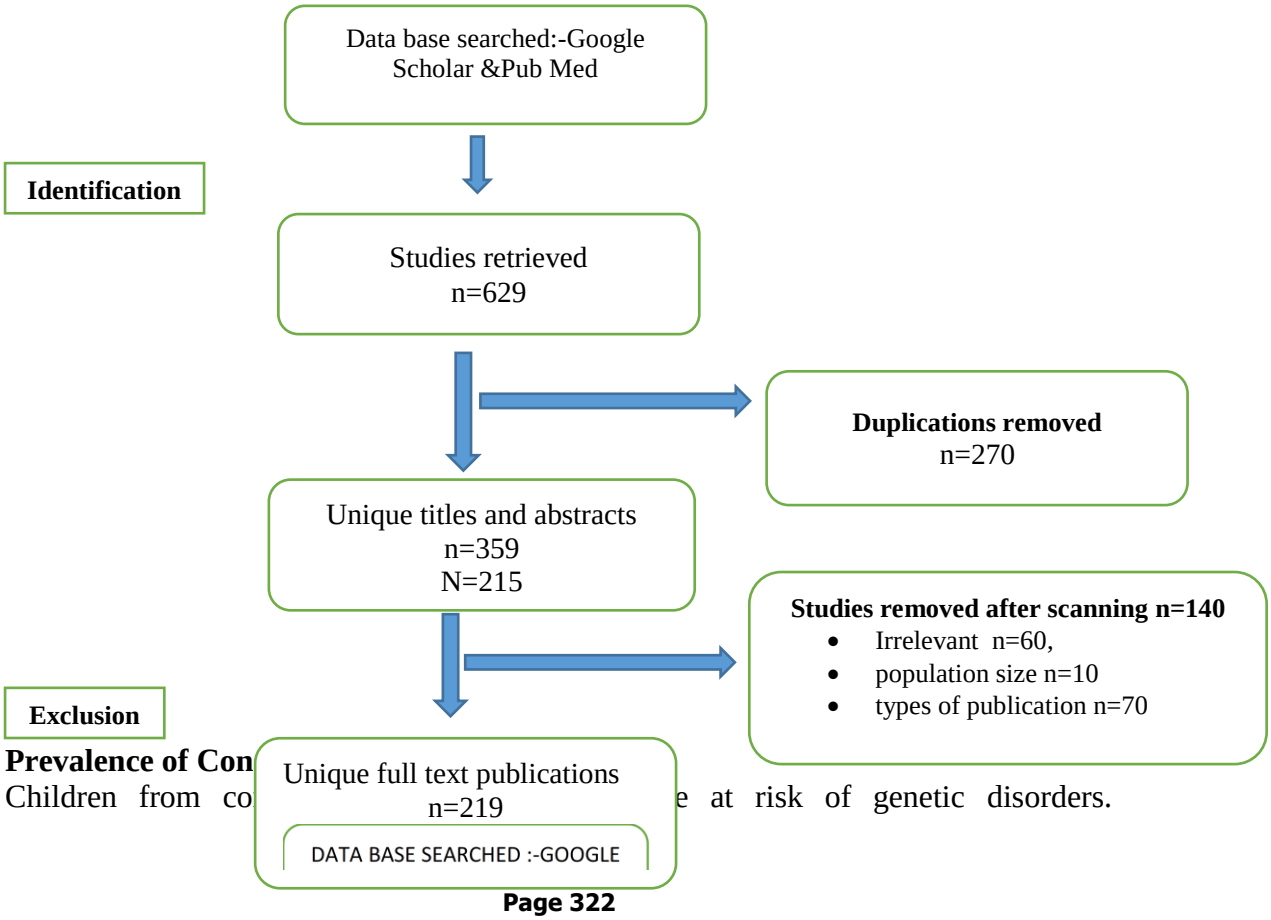
% in 2013 in Pakistan (Manzoor et al., 2018; Hayat et al., 2022).
In this review, we will discuss the prevalence rate of consanguinity and its association with different genetic disorders found in Pakistan. The published research work is selected from the last 10 years (2015 to 2025) of research articles.

Methodology

A comprehensive review was conducted to explore the consanguinity and its association with genetic disorders in Pakistan by using online data sources, "Google Scholar and Pub PubMed ". To explore the breadth of research data available in published work done between the period of 2015 and 2025 by using keywords such as "consanguinity", "genetic disorders", and "Pakistan". Research Studies providing deep insights into genetic abnormalities and their prevalence in Pakistan were preferred. Non-English articles and articles having non-significant data on consanguinity and genetic disorders were not included. Also, the articles that were commentary pieces, were editorials, or lacked empirical data were not added. To ensure the validity and reliability of the review, data were collected from the review journal articles, original research articles, and systematic reviews to explore the consanguinity and genetic disorders in Pakistan. Key data was extracted by reviewing the articles selected, preferably, including population size, geographic locations, prevalence rate, and genetic disabilities. No ethical approval was required because the data were collected from already published work on this topic. However, available data sources were deeply analysed for scientific precision, ethical conformity in the collection of original data, and publications in esteemed journals.



Prevalence of Con
Children from co e at risk of genetic disorders.

Studies excluded n= 135

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Consanguinity has deep social roots in Pakistan. Ratio of consanguinity to non-consanguinity is almost 2 to 1. Consanguinity is more common in families residing in Punjab and Sindh as compared to those residing in Balochistan. Modern age factors, such as an increase in women's educational status, urbanization, and an increase in marriage age, can contribute to the trend of consanguinity in Pakistan. (Kamal et al., 2015). In a cross-sectional study conducted at Islamabad, the prevalence of consanguinity was observed as 62% while 38% had non-consanguineous marriages. The average age of a woman having consanguinity was 21 years, as compared to 24 years for non-consanguineous marriages 18 % women were illiterate in consanguinity, as compared to 4.5% in non-consanguinity (Khan & Mazhar, 2018). Consanguinity is very common in Pakistan, but mostly surveys are performed in urban areas, and the data collected from urban areas is very limited. In a cross-sectional study, consanguinity was observed in 56.72% and the inbreeding coefficient was 0.0348. First cousin marriage rate was very high (49.11% of all marriages), and marriages up to distantly related/Biradari comprised 67.94% of all marriages. The occupation of husbands in consanguineous marriages was found mostly to be landowners (77.59%) and businessmen 62.62%(Hina & Malik, 2015). The survey was conducted in Bhimber district of Mirpur division, Azad Jammu and Kashmir, Pakistan results showed a consanguinity prevalence of 62% of the total marriages (F=0.0348). First-cousin marriages were predominant and formed 50.14 % of all marriages (Jabeen, 2014 #10). The prevalence of consanguinity in four districts of northwest Pakistan: Haripur, Muzaffarabad, Mansehra, and Shangla calculated to be 55%, and the inbreeding coefficient F (ICF) was estimated to be 0.029. The rate of CU was seen to be declining over time, and in marriages that started 'before 1980' and 'after 2010 respectively, ICF from 0.030 to 0.027. Also observed that the literacy rate and marriage age were also increased, and the prevalence of reciprocal marriages decreased over time (Roy et al., 2020). Cross cross-sectional study in seven tehsils at Lower Deer was conducted, rate of consanguinity was observed respectively in Samar Bagh, Timergara Balam bat (57%) (55%), (54%). The rate of consanguinity was higher in rural areas compared to urban areas, 46% and 39% respectively (I. Ahmad et al., 2016). A study on Pashtun family consanguinity prevalence observed 22.34%, also the inbreeding coefficient F was calculated to be 0.0134 (B. Ahmad et al., 2016).

CONSAGUINITY AND NEUROGENETIC DISORDERS

Studied that subjects, 60 % couples having neural tube pregnancy defects were cousins. Of the 60%, there were 46.84 % first cousins. Neural tube disorders anencephaly (18.95%), encephalocele (4.74%), meningocele (42.11%), and myelomeningocele (30%), were diagnosed. Neural tube disorders are multifactorial. Most subjects (63.53%) reside in rural areas (Nauman et al., 2016). Riazuddin et al studied that Intellectual disability (ID) is a heterogeneous genetic disorder, affecting 1–3% of the overall population. Results showed of exome sequencing of 121 consanguineous Pakistani ID families. Out of 121,60 families showed homozygous and heterozygous genetic variants of a single gene, 30 showed already reported ID genes, and 30 families showed novel ID genes (Riazuddin et al., 2017). Saadi et al reported that spinocerebellar disorders are a vast group of rare neurogenesis

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conditions, so nine consanguineous families showing different spinocerebellar phenotypes were analysed using whole exome sequencing. After analysis, 6 novel pathogenic variants were detected in these families, and also reported 3 pathogenic variants were also identified (Saadi et al., 2023). Haider et al investigated inheritance patterns of spinal muscular atrophy (SMA), reviewing the records of tertiary care public hospitals, and reported that 67% of SMA patients showed consanguinity (Haider et al., 2022). Nauman et al discussed that 190 pregnant women coming for delivery to Rawalpindi hospital had neural tube disorders in pregnancy 60 % couples showed consanguinity, out of 60% 46.84 % first cousins (Nauman et al., 2016). Ali et al investigated that Epilepsy is a serious neurological disorder with prevalence in low-income areas of Pakistan cross-sectional study showed demographic and clinical factors, including disease onset, age, gender, and parental consanguinity. Males were found to be more affected, while females were less. Prevalence was high in individuals aged 11 to 20 years. Most of the patients reported the paternal consanguinity (Ali et al., 2023). Ahmed et al studied 5 consanguineous families from Khyber Pakhtunkhwa province of Pakistan were analyzed through genome sequencing, Sanger sequencing, and genetic mapping. And reported an abnormal spindle-like microcephaly gene. Mutation analysis of the gene identified missense mutations and a deletion mutation in all consanguineous families (Ahmed et al., 2019).

CONSAGUINITY AND BLOOD DISORDERS

Akhtar et al reported that Thalassemia syndromes are genetic disorders that show health risks in regions where the consanguinity rate is very high. Cousin marriages are common in Southeast Asia, the Mediterranean, and Africa, especially with a high prevalence rate of up to 73% of β-thalassemia in Pakistan and India. (Akhtar et al., 2025). N. Khalid et al studied 200 participants, including thalassemia patients, out of which males and females were 52% and 48% respectively, and 78 % were relatives, first cousins were 61% and 17.5% were not consanguineous relatives t relatives It was concluded that thalassemia is directly related to consanguinity. (N. Khalid et al., 2019). Aziz et al performed research on patients with thalassemia and observed that 79% of the patients were the result of consanguinity, emphasising the influence of genetic factors in the prevalence of Thalassemia. The prevalence of consanguinity is 86% families in the study group. (Aziz et al., 2024). Tariq et al included 263 people in a study, out of which 111 (42.2 %) were patients. Out of these 111 people with hemoglobinopathies, 86 (32.7%) had minor B-thalassemia and 22 (8.4%) had major B-thalassemia, sickle cell disease was 3 (1.1%). Out of these, 40.3% of patients' parents were cousins (Tariq et al., 2023).

CONSAGUINITY AND INBORN ERRORS IN METABOLISM

Inborn errors of metabolism (IEM) are a heterogeneous group of genetic disorders present in all ethnic groups. Afzal et al reported that the frequency of consanguinity among parents of newborns with IEM diagnosed by neonatal screening was very high in patients of Pakistani, Afghan, Turkish, and Arab areas (71.4%). IEM prevalence was 25.5 times higher in patients of Pakistani, Turkish, Afghan, and Arab areas versus to ethnic Danish children. The prevalence of IEM was 30-fold and 50-fold more in

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Pakistanis (6.5/10,000) and Afghans (10.6/10,000), respectively, versus ethnic Danish children. The results showed a deep link between consanguineous marriages and IEM (Afzal et al., 2018). Cheema et al investigated inherited metabolic diseases in children of a tertiary care hospital, Lahore, Pakistan. Out of 239 patients, 19 types of genetic metabolic disorders were observed in 180 patients. Consanguineous marriages were found in 175 (97%) of the parents of the patient children (Cheema et al., 2016). Hereditary Tyrosinemia Type 1 (HT1), a rare genetic disorder metabolic disorder caused by fumarylacetoacetate (FAH) enzyme deficiency, resulting in toxic metabolite buildup. S. A. Khan et al detected 6 patients, 4 males and 2 females, of HT1 who were a result of consanguineous marriages in their parents. The study highlighted clinical findings of Tyrosinemia Type 1 (HT1) in a cohort of Pakistani children. Also showed challenges in diagnosing, referral, and treatment in Pakistan. HT1 metabolic hereditary disorder is highly associated with consanguineous marriages in Pakistan (S. A. Khan et al., 2024).

CONSAGUINITY AND GENETIC DISORDERS OF THE EYES

Ali et al studied 152 cases of eye genetic disorders. Strabismus was found in 27.6%) patients, cataract (25.6%), myopia (21.1%), microphthalmia (8.5%), exophthalmia in (5.3%), astigmatism in (5.9%) and nystagmus in (3.9%) patients. Keratoconus, glaucoma, and retinitis pigmentosa were found (0.7%). 42 cases with strabismus showed 24 maternal and 18 paternal consanguinities. There were 39 cases of cataract, 22 showed maternal and 17 showed paternal consanguinity. So, 32 cases with myopia showed 21 maternal and 11 paternal consanguinities. Overall all 92 patients showed maternal consanguinity, while 60 patients showed paternal consanguinity (Ali et al., 2016). Akhtar investigated a group of 359 males and 346 females, reporting 19 (5.29%) dichromat males and 2 (0.58%) females. The study group comprised 23 castes. The prevalence of consanguineous marriages in Chenot district of Pakistan was 84.82% and the prevalence of consanguinity in the dichromate families was 85.71%, out of which 52.37%were first cousins (Akhtar, 2019). 50 Pakistani consanguineous families from different provinces had pedigrees showing 50 probands, which were examined through the whole genome sequencing technique, having eye disorders like IRDs, cataract, and structural abnormalities. Four responsible genes for inherited anomalies were found. Mostly, pedigrees showed an autosomal recessive mode of inheritance of eye-related genetic disorders(Zafar et al., 2025). A cross-sectional study was performed in Islamabad to find the consanguinity in patients of refractive errors, strabismus, and keratoconus. The rate of refractive error disorder was found to be 83.6% showing a very high frequency among the three disorders 53% patients found were a result of cousin marriages, mostly first cousin marriages of parents (Rahman et al., 2023). Awan et al conducted survey at Shahida Islam Medical and Dental College Lodhran and Jinnah International Hospital, Abbottabad 53 patients of keratoconus were recruited 31 female and 22 makes,29 (54.7%) patients showed first-cousin marriages in their parents, 16 (30.2%) showed second-cousin weddings, 5 (9.4%) showed third-cousin consanguinity level and 3 (5.7%) showed non consanguineous marriages. |High level of linkage between consanguinity and keratoconus was found in patients at the $p < 0.05$ level. The first level of consanguinity showed the most alarming situation (Awan et al., 2022). At the

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Ophthalmology department of The University of Lahore, 30 patients of keratoconus were studied. Among these patients, 56.7% showed the first level of consanguinity in their parents. While 26.7% patients showed the second level of consanguinity among their parents, 6.7% patients showed the third level of consanguinity among their parents, and 10% patients showed no level of consanguinity among their parents. Consanguinity and KC showed an association with $p< 0.05$, showing the results were significant. Results showed overall results of this study showed that the prevalence of the keratoconus genetic disorder is more severe in the consanguineous population as compared to the rest of the population (Afzal et al., 2023). A study at LRBT Hospital and Mughal Eye Hospital, Lahore, also showed that consanguinity is the most alarming factor for congenital cataract in Pakistan (Saba & Irshad, 2022).

CONSAGUINITY AND MENTAL RETARDNES

A group of mentally retarded persons was recruited in a study, and karyotyping was performed 24 MR persons showed chromosomal aberrations. Out of these 24 abnormal karyotypes, 12 cases showed trisomy having Down's syndrome, showing a high prevalence rate. 5 showed Turner syndrome, and 5 showed mosaic Turner syndrome; only 1showed Klinefelter and Cri du Chat syndrome. The ratio among MR patients observed of consanguine and non-consanguine origin was 61.76 % and 38.24%, respectively. So, finally been proven that consanguinity and intellectual retarders are highly associated. (Hussain et al., 2020). Mental Retardation (MR) is associated with abnormality in intellectual functioning and adaptive skills. Khan et al studied consanguineous families from South Waziristan Agency (SWA) for mental retardnes analysis, each family having at least 2 affected persons. The clinical reports showed abnormalities in patients like short head circumference, abnormal hands and feet, but some MR patients did not show such abnormalities, so the heterogeneity of autosomal recessive MR genes was proved. Finally, it was proved that 50 % of cousin marriages in the region are responsible for mental regardless cases (Khan et al., 2017).

CONSAGUINITY AND CARDIOVASCULAR DISORDERS

Congenital heart disorders and consanguinity are highly associated in the Pakistani community. A study of 14 patients who were born to consanguineous parents showed that cousin marriages are an alarming factor for the offspring in terms of congenital cardiovascular genetic disorders (Fazeriandy et al., 2018). In a study conducted at Faisalabad Institute of Cardiology (FIC), Faisalabad, Pakistan, it was observed that 66.4% of patients with congenital heart disorders have consanguineous Multivariate analysis indicated that consanguineous marriages have a high association with cardiovascular disorders in children, showing an odds ratio of 5.84 (95% CI 4.2-7.99) with a P-value of <0.01 (Mughal et al., 2022). Shah et al studied a group of 346 children on echocardiography at Khyber Teaching Hospital, Peshawar. It was found that 173 children showed a congenital heart disorder. Congenital heart disorders were higher by 1.68 in cousin marriages as compared to non-cousin marriages, with a p-value of 0.02 (Shah et al., 2020). The recessive genetic mode of inheritance is an alarming factor for congenital heart disorders in Asian communities due to the high prevalence rate of consanguinity. This was investigated in a group of 49 CHD probands, collected from 48 consanguineous families in Pakistan. Most of the

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patients (82%) had septal genetic disorders (Baird et al., 2024).

CONSAGUINITY AND DENTAL GENETIC DISORDERS

Abbas et al conducted a study at Foundation University College of Dentistry in Pakistan. 297 participants with congenital anomalies were investigated, prevalence of consanguinity was found in 210 (70.7%) participants. While congenital dental anomalies with consanguinity were found to be significant with p p-value of 0.007 (Abbas et al., 2022). A cross-sectional household survey was conducted using systematic random sampling. 6-9-year-old children and their parents were examined. Results expressed dental anomalies like supernumerary teeth in fathers (p =.009); fusion teeth in mothers (p = 0.002); fusion (p < 0.001), and microdontia (p = 0.002) in respondents. All dental anomalies showed a significant association with consanguinity. (Khan, 2018). Bağcı et evaluated nonsyndromic developmental dental anomalies (DDAs) in patients having consanguineous and non-consanguineous parents. A significant statistical relationship was observed between consanguinity and the size (microdontia) and morphological (dilacerations and taurodontism) types of dental anomalies (Bağcı et al., 2022). Congenital dental anomalies are observed with a high prevalence rate in consanguineous families having lower socioeconomic status. Dental anomalies and offspring of cousin marriages showed a positive correlation, which is either due to recessive harmful gene expression or inheritance of a defective gene (Vasekar et al., 2023).

CONSAGUINITY AND SKELETAL MUSCULAR GENETIC DISORDERS

Bone disorders are among the main health risks in the Pakistani community and are increasing with the passage of time. Round about 72 % Pakistani individuals is facing bone disorders. Females are at more risk. Many reports on bone disorders in Pakistan showed different disorders like osteogenesis imperfecta, aget's disease of bone, spondyloepimetaphyseal dysplasia, etc. The high prevalence of genetic bone anomalies in Pakistan is due to a lack of awareness and cousin marriage. The actual reason behind the high prevalence rate of bone anomalies in Pakistan is due to the high rate of consanguinity (Arshad & Ashiq, 2016). Congenital limb defects (CLD) with a broad spectrum of phenotypes among the basic causes of disabilities. Riaz & Malik conducted cross cross-sectional study at the RYK district of Pakistan and registered 2,204 married women and girls, among them 11 patients of CLD, found a prevalence of 4.9/1000, mostly polydactyl, but with a short prevalence range: brachydactyly, camptodactyly, and oligodacty were also observed. All bone anomalies were associated with parental consanguinity (Riaz & Malik, 2021). Khan et al surveyed hospitals, schools, and villages of the district Bahawalnagar, Punjab, Pakistan selected three consanguineous families for study, having patients of syndactyly. Pedigrees were designed and found the ratio 2:1 of foot and hand syndactyly. All consanguineous families showed a percentage of complete and incomplete syndactyly 50% each. The mode of inheritance of genetic syndactyly was observed in 2 types: autosomal dominant and autosomal recessive (Khan et al., 2020). In a retrospective study at Aga Khan University, all pregnancies diagnosed with fetal skeletal dysplasia were studied via ultrasound. Among all skeletal abnormal babies, 47.8% were the result of consanguinity (Zaidi et al., 2025).

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Duchenne muscular dystrophy (DMD) is a neuromuscular disorder. Aziz et al conducted a retrospective study and observed that consanguinity is highly associated with DMD with p p-value of 0.0021(Aziz et al.). Muscular dystrophy (MD) is an inherited neuromuscular disorder showing muscle weakness, atrophy, and loss of function. Hayyat et al conducted a mixed-epidemiological and clinical study on 280 individuals present in families having MD patients at the Sahiwal division. And observed the prevalence rate of MD 10.25%. 85.71% families sowed consanguineous marriages (Hayyat et al., 2025).

CONSAGUINITY AND CONGENITAL GENETIC DISORDERS

Congenital abnormalities are associated with consanguinity. Consanguineous marriages result in stillbirths and abortions, and 2-4 % of live births have physical, mental, and cosmetic congenital disorders. It was concluded that parents having first cousin marriages are more responsible for the inheritance of congenital abnormalities than parents having second and third cousin marriages (Ahmad et al., 2023). Congenital malformations either at the prenatal stage or after birth were observed in a survey of 200 participant's results showed that 68 % are the result of consanguineous and 32 % non-consanguineous marriages. Hydrocephalus, anencephaly, cleft lip/palate, Ventricular Septal Defect, Atrial Septal Defect, Patent Ductus Arteriosus, and Tetralogy of Fallot are the congenital anomalies that were observed (Gul et al., 2021). In a survey of public and private hospitals in Peshawar, it was observed that parents of 73.5% genetic disorders carrying children had cousin marriages. (4.8%)Children had Down Syndrome, 5.7% had CHD, 82.6% thalassemia, 1.7%had intellectual retardation, and 5.2%had Charcot Tooth Syndrome (Sarwar et al., 2022). S. Khalid et al studied at Okara and observed that consanguinity and congenital disorders are significantly associated. Results showed that congenital disorders at the time of birth had p p-value of 0.002 (S. Khalid et al., 2019). In Rawalpindi, different types of congenital anomalies were observed in children, like neurological disorders (39.1%), neuromuscular disorders (21.1%), limb defects (13.5%), musculoskeletal defects (7.4%), blood disorders (4.3%), orofacial defects (3.9%), metabolic disorders (3.7%), and Others (7.1%). 70% of the affected children showed parental consanguinity. (Shaheen et al., 2023). Nawaz et al conducted cross sectional study survey at public places, door to door and hospital and observed following types of congenital anomalies in participants, Limb defects (21%), neurological disorders (16%), neuromuscular disorders (15%), sensorineural/ear defects (15%), ectodermal defects (9%), musculoskeletal defects (7%), and others (4%). 67% parents showed parental consanguinity (Nawaz et al., 2025). According to a survey conducted in Hyderabad, Sindh, it was observed that birth anomalies were 2 times higher in consanguineous marriages as compared to non-consanguineous marriages. Results showed a 11.3 % prevalence of congenital genetic disorders (Kazi et al., 2020)At Sukkur (Sindh), the prevalence of congenital disorders among consanguineous marriages was 15.6% as compared to non-consanguineous marriages, having a prevalence of 3.7 (Soomro & Tikmani, 2016).

CONSAGUINITY AND GENETIC DISORDERS OF SKIN

Oculocutaneous albinism (OCA) is an autosomal recessive syndromic and non-

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syndromic disorder of the melanin pigment. In Pakistan, autosomal recessive non-syndromic OCA is found and is directly proportional to consanguinity (Ullah, 2022). Epidermolysis bullosa is a genetic disorder found in childhood. It is common in those Pakistani families that have a high rate of cousin marriages (Khan & Tareen, 2021). Xeroderma Pigmentosum is an inherited autosomal recessive genetic disorder. Patients show high sensitivity to UV rays. Larik et al reported five cases from Baluchistan and diagnosed clinically, and complications were noted. It was concluded that XP genetic disorder is associated with consanguinity (Larik et al., 2021). Oculocutaneous albinism recessive genetic disorder, is very common in Pakistan. The basic reason behind this is the marriage's higher prevalence rate in Pakistan, according to research work done on Pakistani albino families, following types: - OCA1, OCA2, OCA3, OCA4, OCA5, OCA6, and OCA7 of Oculocutaneous albinism are found in Pakistan (Shah et al., 2018). Harlequin Ichthyosis (HI) is a dreadful skin disorder. HI shows a recessive autosomal inheritance pattern with a frequency of 1in 300,000 live births in Pakistan, which is increasing gradually due to consanguineous marriages. In Pakistan, a case of HI was reported by Devnani & Kumari, who were born at 36 weeks of gestation from consanguineous parents (Devnani & Kumari, 2019). Fatima et al reported a case of autosomal recessive congenital ichthyosis (ARCI) with cataract, which is a rare phenomenon, and no such case had been reported before in Pakistan. Case was a female child of five years old who was born to consanguineous parents. So concluded that this special case of skin genetic disorder is highly associated with consanguinity (Fatima et al., 2023).

CONSAGUINITY AND INTELLECTUAL DISABILITIES

Reading skills like word identification and reading comprehension of people were evaluated comparison was made between two groups first group containing children of first cousins' parents, while the second group contained children of unrelated parents. Results showed that children of the first group expressed more difficulties as compared to the second group and obtained scores of 62.5 and 78.9, respectively (Talaat, 2024)To evaluate risk factors for Intellectual Disability in Pakistani children, Omar & Kokab conducted a study, there is too significant difference was found between the case and control groups. Consanguinity showed a significant p = 0.00. So proven that Consanguinity is the most negative factor for intellectual disability. So, premarital counselling and health education awareness are necessary for people. (Omar & Kokab, 2019). Consanguinity is a major risk factor for the prevalence of autism spectrum disorder (ASD). Studies have proven that consanguinity is a major risk factor for the prevalence of inheritance of autism spectrum disorder (ASD) (Roy et al., 2020). A survey was conducted at schools of the Lahore region to verify the association of intellectual disability and consanguinity results proved that there is a significant association of intellectual disability with consanguinity, with p-value p=0.001.(H. Khan et al., 2024)In another study comprising 298 participants was also confirmed that intellectual disability has a significant association with consanguinity, with p p-value of p-=0.001. Whole genome sequencing was done on 307 families of participants, and 26 novel genes of intellectual disability were found (Harripaul, 2021).

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CONSAGUINITY AND GENETIC SYNDROMES

Chondroectodermal syndrome or Ellis Van Creveld (EVC) is a very rare genetic disorder in the world and in Pakistan also. The common features are dwarfism, ectodermal dysplasia, bilateral postaxial polydactyl, and congenital heart defects. Only 150 cases have been reported all over the world. Parental consanguinity was found in 30 % cases of this syndrome. So proven that consanguinity is also responsible for EVC syndrome {Jan, 2018 #8}. Yunis-Varon syndrome is an autosomal recessive genetic disorder showing features like facial abnormalities and limb abnormalities. Siddique et al reported a case of Yunis Veron syndrome in Pakistan. This is the first case in Pakistan. A baby born to consanguineous normal parents. This family had also born two babies with this syndrome already, but they died in infancy. So proved that consanguinity is also responsible for Yunis-Veron syndrome {Siddique, 2019 #9. In a cross-sectional study conducted at Peshawar on 230 children having 73.5 % consanguineous parents, among other disorders, 11 (4.8%) children also showed Down syndrome. And 5.5% children showed Charcot-Marie-Tooth syndrome (Sarwar et al., 2022).

CONSAGUNITY AND GENETIC SEQUENCING MODERN TECHNOLOGIES

Several genetic disorders have been investigated through linkage analysis and single-gene sequencing for many years in Pakistan. The research techniques like Sanger sequencing, sequence-tagged sites (STS) markers, linkage analysis, and single-nucleotide polymorphism (SNP) array are very costly and time-consuming when several genes are to be investigated. The next-generation DNA sequencing (NGS) technologies have revolutionized the field of medicine and genetics. Heterogeneous disorders and genes causing complex phenotypes having many mutational coding regions can be investigated by using the whole exome sequencing (WES) tool (Umair et al., 2018). Skeletal dysplasias (SKDs) are a rare genetic disorder of the human skeleton caused by more than 750 genetic mutations. In a study of seven consanguineous families in Pakistan, panel sequencing was performed 22 causative new variants were found, including frame shift, nonsense, and missense variants from various genes causing different types of SKDs, across multiple genes known to cause different types of SKDs. It was concluded that panel sequencing is an effective tool to explore the genetic basis of SKDs present in consanguinity (Kakar et al., 2024). Retinitis pigmentosa (RP) is a functionally losing genetic disorder of the rods and cones in the eye. From Punjab, Pakistan, 2 consanguineous families were investigated. The propends and unaffected persons' exomes were sequenced through next-generation sequencing (NGS) type WES (whole exome sequencing) data were analyzed via different computational tools. Many pathogenic mutations were observed. This study highlights the potential application of WES for a rapid and precise molecular diagnosis for heterogeneous genetic diseases such as RP. So proven that the WES tool is very helpful for the molecular study of severe genetic disorders like RP(Babar et al., 2022). Neuromuscular diseases (NMD) are a group of peripheral nervous system neurological genetic disorders. A study of a group of patients having 83% consanguinity was subjected to genetic testing by NGS techniques, like the gene panels tool and WES tool, on blood samples taken from the peripheral region of the

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body. Many genetic sporadic mutations and variants were discovered successfully. (Al-Hedaithy et al., 2025). In a study of 5 consanguineous families from rural areas of Pakistan, neuromuscular genetic disorders were identified, and whole exome sequencing was performed on blood samples, and novel variants were identified in reported disease-causing genes. So highlighted WES importance as an effective diagnostic tool for persons with neuromuscular disorders in LMICs, the areas having health care problems (Aleem et al., 2025).

A genetic disorder of skull microcephaly, primary hereditary (MCPH), was identified in a large consanguineous Pakistani family. Targeted next-generation DNA sequencing was performed DNA sample from the proband with MCPH to identify genetic sporadic mutations by applying a previously developed panel targeting 46 microcephaly-causing genes. All identified variants were verified through Sanger sequencing. In the results, a novel homozygous nonsense mutation, c.7543C>T, in the ASPM gene was observed. (Khan et al., 2018). Inherited retinal dystrophies (IRDs) are a genetic disorder of the eye. In this disorder, genetic mutations cause anophthalmia and cataracts, causing structural abnormalities in the human eye. In a study, fifty consanguineous unrelated families from various provinces and regions of Pakistan were recruited. Whole exome sequencing (WES) was performed on the proband of all families. The WES tool identified twelve new variants of ten already known IRD genes. These genes were CRB1 14.3%, AIPL1 9.5%, GUCY2D 9.5% and CERKL 7.1% and overall 40.4% of all investigated cases (Zafar et al., 2025).

COMPARATIVE ANALYSIS OF CONSAGUINITY AND ASSOCIATED DEMOGRAPHIC VARIABLES IN DIFFERENT AREAS OF PAKISTAN

Comparative analysis of consanguinity and associated demographic variables in different areas of Pakistan data as shown in the table.1, overall, exposes the reality that the rate of consanguinity in rural areas of Pakistan is higher than compared to urban areas. It also highlights the reality that consanguinity is inversely linked to literacy rate and economic status. The families where the education level, especially for females, is higher, the rate of consanguinity is lower. Families having high economic status are less vulnerable to consanguineous marriages.

Table 1: Comparative Analysis of Consanguinity and associated Demographic variables in different Areas of Pakistan

Study	Setting	Consanguinity prevalence		Residency		Literacy rate		Economic status	
		Consanguineous marriages %	Non-consanguineous marriages %	Rural %	Urban %	Educated %	Uneducated %	High %	Low %
(Nawaz et al., 2021)	Okara 2016-17	61%	39%	65.5%	34.5%	35.1%	64.9%	37.4%	62.6%

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(Riaz et al., 2016)	Distt. (RYK)	58.5%	41.5%	60.7%	54.5%	45.0%	65.6%	50.7%	70.6%
(M. Q. Khan et al., 2024)	Baluchistan	51%	49%	57%	43%	Not found	Not found	44%	56%
(Naqvi et al., 2024)	Peshawar 2017-21	58%	42%	72%	28%	43%	57%	22%	78%
(Mughal et al., 2022)	Faisalabad 2019-20	64.4%	36.6%	58.85 %	41.15 %	Not found	Not found	Not found	Not found
(Afzal et al., 2023)	Abbottabad 2022	84.9%	15.1%	60.03 %	33.97 %	No record	Not found	52.8%	47.2%
(Manzoor et al., 2018)	Sindh 2012-13	71.1%	28.9%	82.4%	52.4%	30.1%	69.9%	56%	75%
(Khan & Mazhar, 2018)	Islamabad	62%	38%	43.3%	70.8%	82%	18%	Not found	Not found

PSYCHOSOCIAL AND ETHICAL IMPLICATIONS OF CONSAGUNITY

Stability and security are basic motivators for consanguinity, but studies have shown that in consanguinity, women often suffer verbal and physical abuse from their male partners. But due to blood bonding tolerance habits rate of women is much higher. It has also been seen that the divorce rate is lower due to the disturbance of the whole family state. The partner selection choice is based on the education and financial status. Illiterate people coming from poor rural areas are much more likely to choose consanguineous marriage to maintain wealth in the family (Popescu et al., 2025). In research from data of 1990 to 2018 was explored the consanguinity and its link with female reproductive health and fertility behaviour in Pakistan were explored. Still, the rate of consanguineous marriages in Pakistan is very high due to cultural, social, and religious beliefs and economic benefits. But married couples are facing many health challenges posing a threat to the mother and child, as well as pregnancy, prenatal and postnatal issues (Iqbal et al., 2022). The findings of research suggested that marriage preferences have strong social and cultural backing that influences major life aspects of individuals. In a study, it was clear that consanguineous preferences are based on social and cultural styles and beliefs. It was also found that there are no Islamic religious teachings which can promote consanguinity; rather, it is appreciated to marry outside the blood relations. This practice may be discouraged through policy-making and awareness campaigns. (Shah & Farooq). There is a low level of literacy, especially in female education high level of fertility and overpopulation is more prevalent in Pakistan as compared to other countries. Cousin marriage rates are high in Pakistan because low economic status creates motivations to cooperate with blood relations, and rates of fertility are high, so more and more cousins are produced, promoting consanguinity in Pakistan (Shenk et al., 2024). A cross-sectional study was carried out at the outpatient departments of the different private and public hospitals. Many patients suffered from the adverse effects of cousin marriage. In a cross-sectional study conducted at private and public level hospitals of Peshawar from November 2019 to April 2020 included patients having severe effects of

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consanguinity in their families were included. The conclusion was that low education, low awareness, poverty, and cultural values had encouraged consanguinity in the Pakistani nation {Sarwar, 2022 #1}. Out of 53 patients of keratoconus having consanguineous parents, 25 (47.2%) cases showed poor socioeconomic family status {Afzal, 2023 #2}.

Pakistan Demographic and Health Survey (2012-13) proved that females having uneducated and low economic status and the least developed regions of the country are more likely to have consanguinity. The prevalence rate of abortions, stillbirths, and miscarriages is very high as compared to non-consanguineous marriages {Manzoor, 2018 #3}. In a study using a convenience sampling technique, a sample of 300 women was selected, out of which 62% showed consanguineous marriages, and 38% showed non-consanguineous marriages. The average age of females presenting for maternal and child health was about 27. Women involved in consanguinity were age of 21 and non-consanguinity were 24 years old. About 18% of women in consanguineous marriages were illiterate as compared to 5.4% of women in non-consanguineous unions. The literacy rate of females having consanguineous marriages was 18% while in non-consanguineous marriages, the literacy rate was observed to be 5.4 % {Manzoor, 2018 #4}.

PUBLIC HEALTH RECOMMENDATIONS

Here we will underscore the challenges related to implementation special focus on health genomics in developing states. Our special focus on Pakistan, which has the world's highest consanguinity rate and prevalence of inherited genetic disorders. Still, Pakistan is facing a shortage of genetic testing, newborn screening programs and health genomic systems. The health care system of Pakistan is facing low funding, despite of burden of infectious diseases may lead to less attention to the genetic health care services. There is a need to step forward in long-term goal setting, resourcing and strategic prioritization {Riaz, 2019 #5}. We have to implement Pakistan's national agenda regarding genetic counselling and testing services. There is an intense need to install NGS (next-generation sequencing) facilities at the local level. Policy-making institutions should pay attention to the training and skill development of genetic counsellors and genetists and institutionalize the public information campaign. Need to consult with world expert organizations, and adoption experiences of successful countries like India and Iran may largely contribute to health genomics. These recommendations may be helpful for Pakistan to overcome the increasing burden of hereditary disorders {Shareef, 2025 #6}. In illiterate and rural areas mostly like consanguineous marriage to save wealth and maintain family status. Due to low levels of education and medical knowledge, people do not have awareness about the negative outcomes of cousin marriages, which in turn increases the rate of infant death, stillbirth and abortion and congenital birth genetic disorders in Pakistan {Popescu, 2025 #7}. In a study of Congenital ophthalmological anomalies, it was observed that the threat of this disorder is more prevalent in developing countries where the rate of consanguinity is higher. Consanguineous marriages were more frequent among parents from the lower middle-income group (80%) compared to the middle (62%) and upper middle-income groups (20%). The rate of consanguinity observed in parents is 80% in the lower middle class, as compared to 60 % in the

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middle class and 20 % in the upper middle class in Pakistan {Abbas, 2025 #8}. In a study, it was explored that Pakistan has a serious burden of thalassemia on society and the healthcare system. This study highlights the need for pre-marital screening and proactive measures for thalassemia I Pakistan to minimize the impact of thalassemia and health care costs. Incorporating successful models of genetic health care from other countries emphasizes an action plan including healthcare system development, policy making and implementation, awareness campaigns, and standard professional training {Ghafor, 2024 #9}.

Low literacy rate and high consanguinity rate urge the need to implement public health policies; findings strongly recommend the need to implement health activities according to the cultural and religious beliefs of the region. The government should tackle public health issues by launching public genetic health programs like premarital genetic counselling and mass screening for carrier populations. Education programs oriented on health belief models should be initiated with a focus on addressing socio-cultural beliefs. Education seminars based on genetic health models should be started, special focusing on religious and cultural beliefs {Khalid, 2019 #10.

FUTURE RESEARCH DIRECTIONS IN PAKISTAN

Consanguinity has long-term impacts on the genetic landscape of Pakistan. In future research, sequencing techniques like WES can be used to explore more and more familial and sporadic variants of genes in consanguineous families. The government should provide modern NGS sequencing facilities at the university level, so that more novel genetic disorders may be researched. Due to a lack of modern genetic facilities at the university level, many genetic disorders are still to be explored in Pakistan. In future mapping of population–specific genetic disorders in Pakistan by huge bio bank projects, carrier frequency studies of familial mutations, use of polygenic risk scores (PRS) to evaluate how consanguinity shapes any disease risk are good platforms for genetic researchers in Pakistan. Rural areas of Pakistan are still ignored regarding genetic research. A lot of genetic research work has to be done because most research work has been done on urban areas only. Data available is insufficient and not up to date, so more statistical population studies, like cross-sectional studies, epidemiological surveys, longitudinal descriptive studies, and cohort studies, are necessary to explore further genetic implications of consanguinity in Pakistan. There is also a need for genetic research on consanguineous families in the socio-cultural and behavioural field. For example, Qualitative studies to explore reasons for consanguinity and barriers to change, to investigate the role of religion, tradition and economics in shaping consanguinity, and intervention studies to investigate how awareness campaigns modify attitudes towards consanguinity. Future research in consanguinity should adopt Policy-oriented research programs like cost-effectiveness analysis of genetic screening of newborn babies and premarital screening projects, Studies on the production of personalized medicines, Studies on public acceptance of government policies (e.g. thalassemia screening program), and Comparative studies with other countries(e.g. Turkey, Iran and Saudi Arabia, etc.) on consanguinity-oriented health programs.

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CONSAGUINITY AND FUTURE CHALLENGES

Consanguineous marriages are practiced commonly in many countries like North Africa, the Middle East, South Asia, Europe and North America, even though it has become illegal practices in many countries. Security and stability are basic factors for consanguinity prevalence, but it has been proven by many studies that women in consanguinity are not safe from physical and verbal torture by husbands. Today, due to increasing urbanization and globalization and education new generation is becoming less receptive for consanguinity due to increasing access of education for women has made women more independent, and easy to fulfill their own life goals not the wishes of family., so we can conclude that despite of some advantages of consanguinity partners should keep the genetic disorder risks in front of eyes and try to save the future of next generations by avoiding consanguineous marriages(Popescu et al., 2025).To analyze a single multigenerational family having genetic disorders is equivalent to the analysis of many small families separately. This strategy attracts the collaborations of international universities for Pakistani researchers. Many DNA samples of consanguineous families are sent out of the country. Sometimes Ph.D. students are also sent along with samples for research collaboration. Universities developed efficient pipelines, whereby students would find families with genetic diseases, extract DNA and carry out linkage analysis, and, in some cases, identify the gene mutations using Sanger sequencing. Many universities in Pakistan have developed genetic labs for DNA extraction, linkage analysis, and finding genetic mutations. So now many publications are made by Pakistani researchers on genetics. However, although good efforts are being made by Pakistan in genetic research, but are limited to pedigree analysis, DNA extraction and low-level mutation finding; modern sequencing and genome analysis techniques are not adopted. There are some reasons for these limitations in Pakistan. Firstly, the cost value given to genetic research projects is very low. Even though some institutions have started the NGS technique, but underutilized still underutilized due to the high cost of consumables and reagents. Secondly, Pakistani researchers feel at ease in becoming coauthors with foreign researchers due to the attractive collaboration facility with foreign universities without bearing the pain of their own laboratory hardships and setup. Lastly, the bureaucratic processes in universities have made it too difficult the availability consumable chemicals in labs and other basic facilities. This makes researchers and faculty members demotivated. While the genetics field in Pakistan has discovered many novel genes, but still these studies has not given any direct immediate benefit to the patients in Pakistani community, So it is necessary to have positive link and encouragement for patients so that patients may rely on results and should also counsel the patient about their future options their future options (Majeed & Mohyuddin, 2021).

CONCLUSION

In this comprehensive review, we have discussed in detail the increasing rate of different genetic disorders in Pakistan and their association with consanguinity. This review has also exposed the increasing risk of autosomal recessive genetic abnormalities, especially congenital anomalies in the offspring of consanguineous parents. It also highlights the role of consanguinity in showing rare genetic syndromes

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and the urgent need for proper genetic counselling and community awareness to manage these risks in society. Overall, the results of different research have shown the sensitivity and future consequences of consanguinity in Pakistan. It also underscores the critical requirement for more genetic research studies in rural areas, also in Pakistan, and highlights the dire need for gene sequencing modern facilities at the university level in Pakistan. It presents future directions of research fields on consanguinity, like the use of NGS technologies for genetic variations, socio-cultural and behavioural field studies, policy-oriented research program studies and interventional studies to investigate the impact of awareness campaigns in shaping consanguinity. This review provides the guidelines for health care policy and practice in the Pakistani community, where consanguinity is very common, also helpful for paediatricians, medical practitioners, genetic counsellors, researchers and social workers.

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