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REVIEW ARTICLE

Community Genetics in Real Time: Congenital Anomalies and Genetic Disorders in an Israeli Arab Town with Elevated Consanguinity Rates

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ABSTRACT

Background: To ascertain rates of congenital malformation and genetic diseases in a local where parental consanguinity is high.

Methods: We reviewed the medical charts of 7,200 children over a 6-year period. The study is a retrospective compilation of the chart reviews in a semi-urban Muslim Arab town which were previously presented.

Results: The results of our study revealed a high rate of congenital anomalies in our study group (7.33%), this compares to other studies in highly consanguineous communities where the frequency of anomalies in children and young adults was found to be 6%.

In our study group, the most important genetic disorders and congenital anomalies (in the group of children whom we studied in this survey) were cardiovascular anomalies, hemoglobinopathies, neural tube defects, monilethrix, and chromosomal disorders.

Conclusions: Our survey, focused on one particular town, is presented as a model of epidemiological ascertainment for other communities as well, and includes recommendations, at a community level, to decrease rates of genetic disorders and malformations. The survey is important considering the numerous worldwide communities and populations where, due to social and religious customs, consanguinity continues to present challenges to efforts by the health system to reduce neonatal morbidity and mortality.

Keywords: Community Genetics, Congenital Anomalies, Genetic Diseases, Consanguineous marriages

1. Introduction

Congenital anomalies and genetic disorders are significant public health concerns, with their prevalence varying across populations due to geographical, ethnic, and socioeconomic factors.[1] It is estimated that up to 30% of all pregnancies may be affected by a morphological error, though many of these result in early spontaneous loss.[2] Among established pregnancies, 3–3.5% of fetuses are diagnosed with congenital anomalies,[3,4] a figure that rises to 4.9–5.1% in our department. While some affected pregnancies result in spontaneous abortion or termination—especially in cases of chromosomal aberrations or multiple congenital defects—others reach term, with approximately 2.7% of liveborn neonates exhibiting congenital anomalies.[3]

Congenital anomalies can be categorized into isolated and multiple anomalies, with approximately one-third of cases presenting with multiple abnormalities. These more complex cases are associated with a higher risk of early mortality, with nearly a quarter of affected children—predominantly those with multiple anomalies—succumbing within the first year of life.[5] Among known risk factors for congenital anomalies, multiple gestation is particularly notable, affecting approximately 8% of fetuses—2.5 times the rate observed in singleton pregnancies. Advanced maternal age further increases the risk, largely due to the higher incidence of chromosomal abnormalities. Additionally, male fetuses demonstrate a slightly higher risk of congenital anomalies, with reported prevalence ranging from 1.3% to 3.5%, compared to a maximum of 3% among female fetuses.[6-8]

Approximately 43% of congenital anomalies are diagnosed at birth, with 82% identified by six months of age. The remaining cases are diagnosed later in life, depending on the nature of the anomaly, the availability of specialized diagnostic facilities, and the expertise of healthcare providers.[9] These factors are particularly relevant when assessing congenital anomalies in specific populations, such as the Arab community in Israel. Arabs constitute nearly 20% of Israel's total population, with approximately 92% identifying as Muslim, while the remaining proportion comprises primarily Christian Arabs and a small Druze population.[10] Notably, a significant percentage of Muslim Arabs are Bedouin, and consanguineous marriage—especially prevalent among Muslims—is a well-established risk factor for congenital anomalies.[11] Recognizing the impact of consanguinity, healthcare authorities have recently implemented targeted public health initiatives, including genetic counseling, prenatal screening programs, and the promotion of folic acid supplementation among women of childbearing age.[12]

Historically, many Arab towns and villages in Israel were founded by a small number of extended families, leading to a high rate of intra-community marriage over generations.[13] This pattern increases the likelihood of genetic founder effects, in which specific hereditary conditions become disproportionately prevalent within a given community. A comprehensive understanding of the genetic landscape in these populations is essential for the development of effective preventive strategies, including targeted genetic counseling and prenatal diagnostic services.

This review aims to examine the incidence and patterns of congenital anomalies and genetic disorders in a representative Arab town in central Israel, home to approximately 35,000 inhabitants. Through an analysis of epidemiological surveys conducted over the years, this study provides critical insights into the burden of genetic disorders in a consanguineous community and highlights the necessity of systematic approaches in diagnosing and managing congenital anomalies in resource-limited settings. The study is also a testament to how primary care physicians, when faced with unexpected epidemiological patterns in their clinical practice, can drive advancements in medical knowledge by systematically documenting, questioning, and collaborating with academic experts. By integrating clinical observations with rigorous epidemiological analysis, this work underscores the importance of equipping healthcare providers with the tools to recognize, investigate, and address genetic disorders in high-risk populations. The findings of this study will contribute to a broader understanding of congenital anomalies in similar populations worldwide and inform strategies for genetic counseling and prevention in communities where consanguinity remains a prevalent practice.

Subjects And Methods

2.1 SUBJECTS

This study examined 7,200 children attending a pediatric center in Taibe over a six-year period between January 1998 and January 2004. The epidemiological data, commentary, and discussion presented in this review are based on previously conducted studies, which were presented at medical conferences and published in conference proceedings over the past two decades. Given that this study is a compilation and review of prior research rather than newly conducted primary research, it does not fall under the standard requirements for ethical review by an Institutional Review Board, which applies to previously unpublished studies.

2.2 MATERIALS AND METHODS

This study is a retrospective literature review of previously published epidemiological studies conducted by the first author. The included studies comprised both prospective and retrospective components. The prospective component included all children born after January 1998, who were examined through routine medical check-ups conducted at scheduled intervals. The retrospective component involved children born before January 1998, with data obtained through medical record reviews. The studies included in this review ranged from October 1991 to October 2014 and encompassed a total of 21 studies: 2 case series, 3 national surveys, 4 cross-sectional studies, 1 genetic linkage study, 1 prospective cohort study, and 10 retrospective cohort studies. Diagnoses documented in medical records were verified through clinical re-evaluation, in which affected children were invited for an examination to confirm the presence of congenital anomalies or genetic disorders. The cutoff age for inclusion was 15 years.To ensure consistency in case identification, only infants or children

with a gestational age of ≥28 weeks or a birth weight of ≥1,000 g were included in the analysis. The study focused exclusively on anomalies diagnosed after the 28th week of gestation that, in the absence of intervention, would have had a detrimental impact on physical function or social acceptance.

Data collection included information on the frequency of congenital anomalies among siblings of the index cases, obtained through parental interviews. While data on perinatal mortality, miscarriage, and intrauterine deaths were recorded, only major anomalies in live-born children were included in the final analysis due to incomplete documentation of fetal losses. All congenital anomalies were diagnosed and confirmed by a pediatrician (L.J.) with extensive experience in identifying major congenital defects. Minor anomalies were noted but were excluded from the primary analysis. After clinical assessment, a complete pedigree and family history were obtained for each affected child to investigate potential hereditary patterns.

Each child with an anomaly was classified according to whether they had an isolated abnormality affecting a single body site (Group 1) or multiple abnormalities affecting more than one body site (Group 2). Children in Group 2 were further classified based on the pattern of anomalies, including syndromes, sequences, associations, complexes, and uncharacterized multiple defect patterns.

3. Results

During the six-year period between January 1998 and January 2004, we identified a total of 528 children with major congenital anomalies, indicating a prevalence of 73.3 per 1,000 live births for the combined prospective and retrospective groups. This figure does not include stillbirths or deaths that occurred prior to the commencement of the study. Of these 528 children, 69 (13.1%) had multiple congenital abnormalities, 416 (78.8%) had isolated abnormalities, and 43 (8.1%) had metabolic or other disorders (Table 1).

Table 1: Prevalence of Major Congenital Abnormalities In 7,200 Children in an Arab Town

Type		No. of Patients	Total
Multiple System	Non Syndromic Syndrome Chromosomal Autosomal Recessive Autosomal Dominant Unknown	23 20 17 0 0 0	69
Single System	Cardiovascular Central Nervous System Genitourinary Skin Ophthalmological Gastrointestinal Musculoskeletal Hematological Ear, Nose and Throat Respiratory	106 90 76 29 43 25 21 14 8 4	416
Metabolic diseases and others	Congenital Hypothyroidism Deficiency of Complex of Respiratory Chain Growth Hormone Deficiency Ambiguous Genitalia Nesidioblastosis Cystic Fibrosis Phenylketonuria Neurofibromatosis Galactosemia Familial Mediterranean fever	5 2 4 1 1 1 1 1 1 3	32
	Familial Hypercholesterolemia Marfan Syndrome Niemann-Pick Disease Pierre Robin Syndrome Hyperinsulinemia Hyperammonemia Hypoglycemia Oxalosis	4 1 1 1 0 0 1 1	

3.1 CARDIOVASCULAR SYSTEM

Anomalies of the cardiovascular system were the most common, comprising 20.1% of the total anomalies (106 out of 528 children). This figure does not include cardiac anomalies that were part of a multiple anomaly condition. Diagnoses were based on clinical examination, echocardiography, and additional investigations. The most frequently observed cardiac anomaly was ventricular septal defect (VSD), identified in 34 cases (32.1% of all cardiac anomalies). The majority of cardiac anomalies were acyanotic, except for 9 cases (8.4%), which were cyanotic.

3.2 CENTRAL NERVOUS SYSTEM (CNS)

We identified 85 cases of abnormalities in the central nervous system (CNS) (16.1% of the total anomalies). Of these, 38 children had developmental delay, 20 had convulsive disorders, 7 had neural tube defects (1;1,000 infants), and five had craniosynostosis that required surgical intervention in the first few months of life (Table 2). 11

Table 2: Single System Involvement among Infants and Children with Major Malformations

Type		No. of Patients	Total
Cardiovascular	Ventricular Septal Defect, (VSD)	34	106
	Pulmonary Stenosis (PS)	15	
	Patent Ductus Arteriosus (PDA)	8	
	Atrial Septal Defect (ASD)	9	
	Mitral Valve Prolapse	6	
	Atrioventrical Canal Defect,	4	
	Ttralogy of Fallot	4	
	Pulmonary Dilatation	3	
	VSD + ASD	3	
	Transient Global Amnesia	3	
	Aortic Stenosis,	4	
	VSD +PS	2	
	PDA+PS	2	
	Single Atrium+ Single Ventricle	1	
	Mitral. Stenosis		
	Coarctation of Aorta,	1	
	Situs Inversus	1	
	Ebstein Anomaly	1	
	Tricuspid Valve,	1	
	ASD + Pulmonary Valve Disease.	1	
	Aortic Valve Insufficiency Total anomalous venous return, Bicuspid aortic valve 2.	1 2	
Central Neurological Anomalies	Convulsions	20	29
	Developmental Delay	38	
	Neural Tube Defect	7	
	Craniosynostosis,	5	
	Mental Retardation	4	
	Microcephalus + MR	3	
	Hydrocephalus	3	
	Peripheral Sensory Neuropathy	3	
	Microcephaly	2	
	Dandy Walker Syndrome	1	
	Agenesis of Corpus Callosum	1	
	Brain Infarction (Thrombophilia) Choreoathetosis	1	
	Convulsions.	1 1	
Genitourinary Anomalies	Hydronephrosis	32	67
	Undescended testis	17	
	Double Collecting System	6	
	Vesico-ureteric reflux,	4	
	Dysplastic Kidney,	3	
	Hypoplastic Kidney	2	
	Ectopic Kidney	4	
	Bifida Collecting System	1	
	Multicystic Kidney	1	
	Cystic Kidney	1	
	Pelviureteric Junction Stenosis	3	
	Single kidney Hypospadias	1 1	

Musculoskeletal Anomalies	Pes Adductus Congenitus, Congenital diaphragmatic hernia, Osteogenesis Imperfecta, Club Foot Syndactyly Arthrogryposis Femoral Torsion Chondrodystrophy Phocomelia Small finger.	4 4 2 2 3 2 1 2 1 0	32
Gastrointestinal	Cleft Lip / Cleft Palate Bifid uvula Syndrome Hirschsprung's Disease Duodenal Atresia, Intestinal Atresia Imperforate Anus Ant Anus Intrahepatic Cholestasis	11 1 4 3 1 1 1 3	32
Respiratory			4
Eye Anomalies	Strabismus Cataract + Strabismus Cataract Retinitis Pigmentosa Anophthalmia Optic Atrophy + Glaucoma Keratoconus Hypemetropia, Macrocornea .	27 3 3 5 1 1 1 1 1	42
Dermatological Anomalies	Monolithrix Epidermolysis Bullosa Cutis Marmorata Congenita Venous Malformation Dermal Nevus Nevus Flammeus / Big Big Hemangioma Dermal Sinus	20 3 0 1 1 1 1 1	32
	Fat Necrosis	1	
Ear , Nose And Throat	Congenital Subglottic Stenosis Microtia Deafness Tracheomalacia Lacrimal Stenosis	1 1 4 1 1	8
Hematological	Thrombastenia, B12 Deficiency, Protein Deficiency Thalassemia HG Taybe Disease Sickle Cell Anemia Hereditary Persistence of Fetal Hemoglobin Anemia	1 3 3 1 4 1 1	24

3.3 GENITOURINARY ANOMALIES

Genitourinary anomalies were diagnosed in 76 children (14.4%). The most common abnormality was hydronephrosis, found in 32 children, followed by undescended testes (n=17). Other anomalies included vesicoureteric reflux (n=4) and double collecting system (n=6) (Table 2).

3.4 OPHTHALMOLOGICAL ANOMALIES

A total of 43 children (8.1%) had ophthalmological anomalies. The most frequent condition was strabismus (n=27, 62.8%), followed by congenital cataract (n=8, 18.6%), and retinitis pigmentosa (n=5, 11.6%) (Table 2).

3.5 DERMATOLOGICAL ABNORMALITIES

A total of 29 children (5.5%) were diagnosed with dermatological abnormalities. The most prevalent condition was monilethrix (n=20, 69.0%), followed by epidermolysis bullosa (n=3, 10.3%) (Table 2).

3.6 MUSCULOSKELETAL ANOMALIES

A total of 21 children (4.0%) had musculoskeletal anomalies. Correction: Congenital dislocation of the hip (previously included as an anomaly) was removed due to its classification as a deformation rather than a malformation. The most frequently observed conditions

were craniosynostosis (n=5, 23.8%), syndactyly (n=3, 14.3%), and clubfoot (n=2, 9.5%).

3.7 EAR NOSE AND THROAT ANOMALIES

Eight children (1.5%) had ENT anomalies. The most common was congenital deafness (n=4, 50%), followed by tracheomalacia (n=1) and congenital subglottic stenosis (n=1) (Table 2).

3.8 GASTROINTESTINAL ANOMALIES

Gastrointestinal anomalies were diagnosed in 25 children (4.7%). The most common were cleft lip/cleft palate (n=11, 44.0%), followed by Hirschsprung disease (n=4, 16.0%), and duodenal atresia (n=3, 12.0%) (Table 2).

3.9 HEMATOLOGICAL AND IMMUNOLOGICAL DISORDERS

Fourteen children had hematological or immunological disorders, including thalassemia (n=4), sickle cell anemia (n=1), and hereditary persistence of fetal hemoglobin (n=1)

3.10 MULTIPLE ABNORMALITIES

A total of 69 children (13.1%) had multiple congenital abnormalities, of whom 12 had recognized syndrome (Table 3). The most common autosomal recessive syndromes included Walker-Warburg syndrome (n=2), mitochondrial depletion syndrome (n=3), and hypertrophic cardiomyopathy with hypotonia and umbilical hernia (n=1).

Table 3: Genetic Contribution

Type		No. of Patients	Total
Autosomal Recssive	Retinitis Pigmentosa Peripheral Sensory Neuropathy Congenital Hypothyriedism Epidermolysis Bullosa Deafness B12 Deficiency Protein C Deficiency Thalassemia HB Taybe Sickle Cell Anemia Hereditary Persistence of Fetal Hemoglobin Anemia Severe Combined Immunodeficiency Factor 7 Deficiency Arthrogryposis Alstrom Disease, Walker-Warburg Syndrome Mitochondrial Depletion Deficiency of Complex of Respiratory Chain Growth Hormone Deficiency Ambiguons Genetalia Nesidioblastosis Cystic Fibrosis Methemoglobinemia Galactosemia Phenylketonuria Intralepatic Cholestasis Familial Mediterranean Fever Mental Retardation Niemann-Pick Disease, Ornithine Transporter Defect VUR grade 2 DiGeorge Syndrome Rett Syndrome + Nystagmus Oxalosis	2 3 5 3 4 3 3 1 5 1 1 3 1 2 1 2 3 2 4 1 1 1 1 1 1 5 3 2 1 1 1 1 1 1 1	72
Autosomal Dominant	Monilitrix Chondrodystrophy, Osteogenesis Imperfecta Rubenstein Taybe Syndrome Osler–Weber Syndrome Pulmonary Arteriovenous fistula Neurofibromatosis Glanzmann thrombasthenia Familial Hypercholesterolemia Marfan Syndrome.	20 2 2 1 1 1 1 2 4 1	22

X-Linked	Hypopigmentosis In Ito Hemophilia	1 1	2
Unknown	Cutis Marmorata Congenital Venous Malformation Hypoplastic Left Heart Syndrome Dermal Sinus	1 1 1	2
Sporadic	Caylor Syndrome Vesicoureteral Reflux Septo-Optic Dysplasia	1 1 1	2
Chromosomal	Down Syndrome, 47 XXY Syndrome Turner Syndrome Velo Cardio Facial Syndrome .	13 1 1 2	26

3.11 UNIDENTIFIED MULTIPLE MALFORMATIONS
Twenty-seven children had abnormalities that could not be classified, either because the investigations were incomplete, or the abnormalities did not conform to any disorder, or the syndrome was unknown to the authors (Table
2).

3.12 GENETIC CONTRIBUTION TO CONGENITAL ABNORMALITIES
Among the 528 children, 123 (23.3%) were suspected of having a single-gene disorder based on clinical diagnosis and inheritance patterns (Table 3). These included 72 cases (13.6%) of autosomal recessive inheritance, 34 cases (6.4%) of autosomal dominant inheritance, and 2 cases (0.4%) of X-linked inheritance.

3.13 PREVENTABLE ABNORMALITIES
Among the identified cases, 103 children with cardiovascular anomalies could have been diagnosed antenatally through fetal echocardiography. Additionally, 22 cases of Down syndrome could have been detected through first-trimester screening and confirmatory genetic testing. Neural tube defects (n=7) were also preventable through maternal folic acid supplementation and early prenatal screening.

4. Discussion

This long-term, community-based study demonstrates a high prevalence of congenital anomalies and genetic disorders in a semi-urban Arab town in central Israel, where consanguineous marriages remain relatively common. Our findings, derived from 7,200 children, reveal a congenital anomaly rate of 7.33%, which exceeds rates reported in both Western and regional literature. Cardiovascular, urogenital, and central nervous system anomalies were among the most common findings, with several rare autosomal recessive disorders also identified.

The overall rate of congenital anomalies in our survey (7.33%) surpasses the 6% prevalence reported in a U.S. study of individuals up to 25 years of age. [13] The consanguinity rate in the study population (22%) was significantly elevated among parents of affected children (45%), reinforcing the established association between consanguineous marriages and autosomal recessive conditions. Similar trends have been reported in other Middle Eastern countries, with consanguinity rates ranging from 25% in Lebanon to over 55% in Saudi Arabia and Kuwait. [14–16]

Cardiovascular, urogenital, and central nervous system anomalies each accounted for approximately 14% of all cases in our study. Among cardiovascular anomalies, ventricular septal defect was the most common, comprising about 25% of all cardiac defects. Atrioventricular septal defects were the most frequent lesion observed in syndromic congenital heart disease, especially in children with Down syndrome, consistent with previous literature. [17,18]

Interestingly, certain autosomal recessive disorders frequently encountered in other Israeli Arab communities—such as non-syndromic deafness and cystic fibrosis—were rare in this population. The prevalence of congenital deafness was 0.56 per 1,000 children, notably lower than the 7.8 per 1,000 reported in another Arab village in northern Israel, where mutations in connexin 26 are common. [17,18] Similarly, only one case of cystic fibrosis was documented in this study, contrasting with the higher frequencies observed in other founder populations.[19,20] These findings suggest the influence of unique founder effects and localized genetic drift within this particular town.[21]

A total of 39 genetic disorders were identified, including 27 autosomal recessive, 9 autosomal dominant, and 3 X-linked conditions, affecting 121 children in total. While clinical diagnosis served as the primary means of identification, many of these disorders clustered within extended families, supporting an inherited genetic basis.

Limitations and Context of the Study

This study represents the culmination of years of dedicated clinical observation and epidemiological tracking in a resource-limited community. During the data collection period (1998–2004), advanced genetic testing, molecular diagnostics, and even some basic biochemical screenings were not consistently available within the local healthcare infrastructure. As a result, diagnoses were made primarily on clinical grounds, guided by the experience of the lead physician (L.J.), who relied on pattern recognition, familial recurrence, and long-term clinical engagement.

Although this approach lacks the precision of genetic confirmation, it reflects the practical reality in many underserved settings where physicians must rely on clinical acumen and epidemiological patterns to identify genetic conditions. While it is possible that some anomalies resulted from non-genetic causes—such as maternal nutritional deficiencies or perinatal infections—

the repeated familial clustering observed across several conditions strongly suggests a genetic origin. These diagnostic limitations, while significant, also underscore the value of longitudinal, community-based primary care as a source of rich epidemiological insight.[22]

Among the identified cases, 103 children with cardiovascular anomalies might have been diagnosed prenatally via fetal echocardiography. Likewise, 22 children with Down syndrome could potentially have been detected through first-trimester screening and confirmatory genetic testing. Neural tube defects (n=7) were also largely preventable through maternal folic acid supplementation.[23]

These findings emphasize the critical need for broader public health interventions aimed at prevention and early diagnosis. Community-wide folic acid awareness campaigns, routine genetic counseling for couples of reproductive age, and the implementation of pre-marital and prenatal screening programs are essential for reducing the burden of congenital disorders in consanguineous populations.[24] While Israeli health authorities have made efforts to promote folic acid use,[10] the uptake remains inconsistent—particularly in minority and rural populations where educational outreach and access to prenatal services are limited.[25]

This study also illustrates the indispensable role that community pediatricians and primary care providers can play in generating valuable epidemiological data. In settings lacking comprehensive hospital registries, physicians in local clinics often serve not only as frontline caregivers but also as primary observers of disease patterns. The systematic documentation of congenital anomalies and inheritance trends over several years enabled the early recognition of rare syndromes even in the absence of laboratory diagnostics.

This grassroots model of data collection demonstrates how clinical vigilance and community familiarity can substitute, to an extent, for formal diagnostic infrastructure. Such an approach offers a valuable framework for identifying public health priorities in low-resource or culturally distinct environments,[22] and may be particularly relevant in similar populations facing healthcare disparities and diagnostic barriers.[26]

Although centered on one Arab town, the findings of this study have broader relevance for consanguineous communities globally. Elevated rates of autosomal recessive conditions are common in populations across North Africa, the Middle East, and South Asia.[27] These parallels emphasize the importance of culturally tailored interventions that both respect traditional practices and work to reduce genetic risks.

Moving forward, efforts should focus on integrating molecular diagnostics and genetic testing into primary care systems, especially in under-resourced areas. Establishing local biobanks and rare disease registries will help support population-specific research and guide preventive strategies. In parallel, expanding the availability of community genetic services and equipping primary care providers with genetics training will enhance early diagnosis and long-term management. Collaborative research networks that facilitate knowledge sharing and cross-regional comparisons will be essential in shaping sustainable and effective models of care.[28–31]

Together, these measures are critical to reducing the incidence of congenital and genetic disorders in high-risk communities and improving outcomes for future generations.

Conclusions:

This study reveals a high prevalence of congenital anomalies and genetic disorders in an Arab town with elevated consanguinity rates, reaffirming the link between consanguinity and autosomal recessive conditions. It underscores the value of community-based clinical research in identifying hereditary patterns and informing public health strategies. The findings highlight the urgent need for improved genetic counseling, routine carrier screening, and expanded prenatal care. Although locally focused, the study's implications extend to other consanguineous populations globally, emphasizing the importance of culturally sensitive preventive programs and multidisciplinary collaboration to reduce the burden of genetic disorders.

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