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Consanguinity, complex diseases and congenital disabilities in the Souss population (Southern Morocco): a cross-sectional survey

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Abstract

Background Several studies showed that the perpetuation of consanguinity increases homozygosity and homogenizes the population's gene pool. This allows the expression of recessive deleterious mutations and increases the prevalence of genetic disorders and birth defects. Despite the reported negative health effects, consanguinity is still practiced in Morocco. This study aimed to evaluate the prevalence and socio-demographic determinants of consanguinity in the Souss region and to assess the association of this type of union with congenital disabilities and complex diseases. To meet this aim, a survey based on a cross-sectional approach was conducted between January 2019 and January 2020 among 520 randomly selected participants in the Souss region.

Results The findings revealed a high prevalence of consanguinity of 28.46%, with first-cousin unions accounting for 16.15% of all marriages. According to multivariate logistic regression analysis, early age at first marriage, men's occupation, endogamy, and parental consanguinity were predictive factors for consanguineous unions in the study population. Moreover, the results revealed a significant association between consanguinity and the incidence of physical disabilities (OR = 3.753; [95% CI 1.398–10.074]), mental retardation (OR = 5.219; [95% CI 1.545–17.631]), deafness-mutism (OR = 4.262; [95% CI 1.004–18.089]) and cardiovascular diseases (OR = 2.167; [95% CI 1.036–4.530]). However, no significant association was found between consanguinity and diabetes, cancer, asthma, epilepsy, and psychiatric disorders.

Conclusion Overall, our results suggest a high practice of consanguinity in the Souss population, associated with social, economic, and cultural factors. Consanguineous unions were associated with a high incidence of mental retardation, physical disabilities, deafness-mutism, and cardiovascular diseases. In this population, where marriage between relatives is highly preferred, awareness programs are not sufficient, and genetic studies on consanguinity-related diseases are necessary to provide specific premarital screening and thus increase the efficiency of genetic counseling.

Keywords Souss, Genetic counseling, Consanguinity, Morocco, Endogamy, Congenital disabilities, Complex diseases

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Background

Consanguinity is a particular pattern of matrimonial behavior consisting of the union between two closely related individuals. From a medical genetics perspective, consanguinity is defined as a union between second cousins or closer with an inbreeding coefficient greater than or equal to 0.0156 [1].

Consanguinity is practiced by up to 10% of the global population, with rates ranging from 80.6% in certain Middle Eastern provinces to less than 1% in Western societies [2]. The prevalence of consanguineous unions differ by region and is fundamentally conditioned by several socio-demographic factors, including social status, education level, religious affiliation, size and location of the area, rural–urban environment, and parental consanguinity [3–5].

Since the twentieth century, increasing industrialization, improved communications, and urbanization have led to an overall decrease in consanguinity [6]. However, an opposite trend was observed in some Middle Eastern and North African countries where consanguinity accounted for 20–52% of total marriages [7].

The long-term practice of consanguinity leads to the homogenization of the population's gene pool. As a result, individuals accumulate homologous chromosomal sequences, increasing homozygosity and, consequently, the expression of some recessive genes [8]. Moreover, as many recessive alleles in populations are harmful to health, consanguinity increases the risk of mortality and congenital, chronic, and infectious diseases [9].

These conclusions are supported by extensive epidemiological studies that highlight the significant influence of consanguinity on children's health, especially within highly consanguineous communities [5]. Thus, associations between consanguinity and a wide range of conditions and diseases have been well-established. Notable examples include Intellectual Disability [10], congenital anomalies [11], non-communicable diseases [12], genetic predisposition to infectious diseases [13], and primary immunodeficiencies [14].

Moreover, the impact of consanguinity on survival has been reported in various studies, highlighting its association with increased progeny mortality [15] and the occurrence of spontaneous abortions [16].

Despite multiple studies documenting the frequency of consanguineous marriages in Morocco, the southern populations have not received sufficient attention. The few studies that have been conducted were based on a single center (sampling site) and focused on a limited number of matrimonial determinants. We attempt to fill this gap in our study by estimating the prevalence of consanguinity in the Souss region, its evolution, and its interaction with socio-demographic factors. Unlike prior research, we collected data from 13 tribes and introduced additional determinants of consanguineous unions, including geographical factors (tribal and village endogamy), inheritance, and the evolution of consanguinity. We also assessed the effects of consanguinity on complex diseases and congenital disabilities.

Methods

Study population

The Souss region was historically part of the Souss Al-Aqsa, which extended from the Mediterranean coasts to the Saharan border before becoming the territory of the Berber tribes in Southern Morocco [17]. The Current Souss region refers to the 46,640.7 km^2 territory surrounded by the High Atlas to the north, the Sahara to the south, the Atlantic to the west, and the Algerian border to the east [18, 19]. The region was officially part of the South Economic Zone until 1997, when it was separated into two administrative zones [20]. Currently, the region includes seven provinces and prefectures belonging to two administrative zones, notably Souss-Massa (Chtouka-Ait Baha, Taroudant, Tata, Tiznit, Agadir-Ida Outanane, and Inezgane-Ait Melloul) and Guelmim Oued Noun (Sidi Ifni) (Fig. 1).

In terms of origin, ancient demographic events have created a diverse population mainly composed of Berber, Arab and sub-Saharan African elements [21, 22]. In addition to ethnic diversity, several cultural and social features characterize this population, such as tribal solidarity and matrimonial traditions.

Study design and collection data

Study design

A cross-sectional study was conducted in the Souss region between January 2019 and January 2020 to determine the impact of consanguineous marriages on offspring health.

Sampling method

Based on the administrative division of the region, a two-stage stratified sampling design was carried out. Due to limited access to the provinces of Chtouka-Ait Baha and Taroudant, only five prefectures and provinces were surveyed (Sidi Ifni, Tata, Tiznit, Agadir-Ida Outanane, and Inezgane-Ait Melloul). First, the number of tribes in each province and prefecture was defined. It was found that there are 45 tribes in total (12 in Sidi Ifni, eight in Tata, 21 in Tiznit, three in Agadir-Ida Outanane, and one in Inezgane-Ait Melloul). Subsequently, it was decided to select a sample of 25% of the tribes, giving equal representation to each administrative region (three tribes from Sidi Ifni, two tribes from Tata, six tribes from Tiznit, one tribe from Agadir-Ida Outanane, and one tribe from

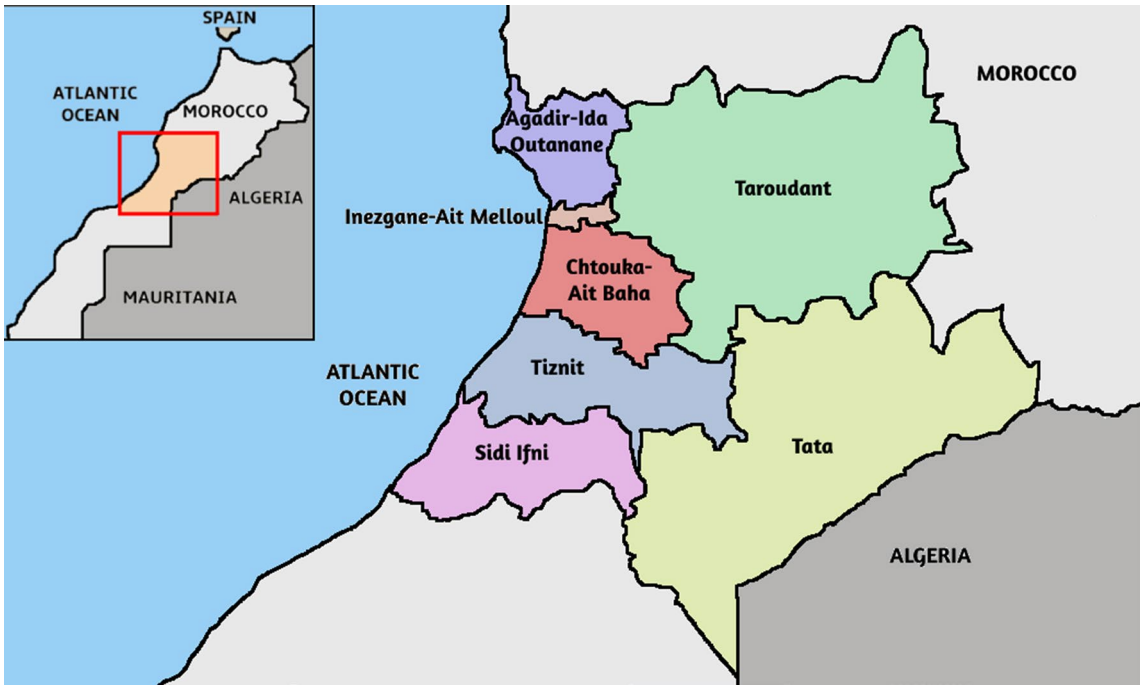

Fig. 1 Administrative organization of the Souss region (prefectures and provinces)

Inezgane-Ait Melloul). Finally, 50 individuals were randomly selected from each tribe for face-to-face interviews, making an initial sample size of 650. However, only 520 individuals agreed to participate, while the remaining subjects were excluded because some refused to participate and others provided incomplete questionnaires.

Subject recruitment

We recruited a total of 520 participants (200 men and 320 women) who gave informed consent to participate in this study. We selected these participants according to the following criteria: ever-married individuals aged 18 years or older and residing within the study region. We excluded individuals under 18 years of age, those who had never been married and temporary visitors to the study region.

Data collection method

The subjects were approached through a house-to-house survey. During face-to-face interviews in the local languages (Arabic-Berber), participants were asked to provide information on their own and their partner's socio-demographic characteristics, such as degree of education, occupation, place of birth (tribe and village/town), age at first marriage, birth environment (urban/rural), and first marriage contraction year. The degree of relationship between couples was noted, as well as whether the participants' parents were related to each other. Data were also collected on the occurrence of diseases and disabilities among the participants' offspring, including complex diseases (cardiovascular diseases, diabetes, cancer, asthma and psychiatric disorders), congenital disabilities (mental retardation, deaf-mute, blindness, physical disability), and infectious diseases.

In order to minimize misreporting and omissions, we combined collective (familial) and individual interviews. In addition, the household interview gave us the opportunity to access medical resources, including family health records, prescriptions and medication orders.

Data analysis

To assess population attitudes toward consanguinity, we calculated consanguinity rates and inbreeding coefficients. We divided unions into six categories according to the degree of consanguinity: Double first cousins ($F=0.125$), first cousins ($F=0.0625$), first cousins once removed ($F=0.0313$), Second cousins ($F=0.0156$), Distant cousins ($F<0.0156$), and no relation ($F=0$). The mean inbreeding coefficient (σ) was obtained by applying the formula [23]:

$$\sigma = \Sigma PiFi$$

Pi is the proportion of each category of consanguineous marriage and Fi is the corresponding inbreeding coefficient.

The consanguinity coefficient includes unions between second cousins or closer ($F \geq 0.0156$). This limit is explained by the low biological effect of an inbreeding coefficient below 0.0156, which is comparable to that observed in the general population [9].

The association between socio-demographic variables and consanguinity was measured using a standard χ^2 test for categorical data and an independent samples *t*-test for continuous-level data.

Adjusted Odds Ratios (OR), along with their corresponding 95% confidence intervals (CI), were calculated for each predictor variable using multivariate logistic regression analysis. This analysis tested the association of each factor with consanguinity while controlling for the effects of other predictors. Odds ratios (95% confidence intervals) were also calculated for the likelihood of diseases by consanguinity status in the participant's offspring. *p*-values less than 0.05 were considered statistically significant.

Results

Prevalence of consanguinity in the Souss region

The prevalence, coefficients and types of consanguinity in the Souss population are presented in Table 1. The global rate of consanguinity was 28.46%, with a mean Inbreeding coefficient of 0.0117. First cousin unions are the most common type of consanguineous unions, which constituted 16.15% of all marriages. Also, the parallel patrilateral pattern (Type I) was the most common in first cousin unions, representing 6.15% of all marriages.

Socio-demographic determinants of consanguineous marriages

Table 2 summarizes the socio-demographic determinants associated with matrimonial behavior among the Souss population. A statistically significant difference was found between consanguineous and non-consanguineous couples according to age at first marriage, men's education, men's occupation, geographical endogamy, and parental consanguinity. For women, consanguinity is more frequent in marriages contracted below the age of 23 ($p < 0.011$). The average age at first marriage is significantly lower ($p < 0.001$) in consanguineous unions than in non-consanguineous unions, being 19.84 ± 0.380 and 21.34 ± 0.317, respectively. For men, age at first marriage between 23 and 28 is the most frequent in consanguineous unions ($p < 0.001$). The average age at first marriage in consanguineous unions is 27.16 ± 0.514, while it is 29.81 ± 0.487 for non-consanguineous unions ($p < 0.001$).

Regarding the birth environment (urban versus rural), for both genders, there is no statistically significant difference between consanguineous and non-consanguineous unions ($p > 0.05$). This result reflects the predominant rural origin of the studied population.

Men's education level was significantly associated with consanguinity ($p < 0.001$). Primary and Quranic pre-school educations were the most frequent among consanguineous unions. For women, there was no statistically significant association between consanguinity and education level ($p > 0.05$). Actually, illiteracy is the predominant feature in the study population, especially among women (421 out of 520 women).

Table 1 Distribution of different types of marriages in the Souss population

Degree of consanguinity	*N*	%	Individual Inbreeding coefficient	Mean Inbreeding coefficient
Non-Consanguineous unions	372	71.54	0	0
Consanguineous unions	148	28.46		
Double first cousins	1	0.19	0.125	0.000240385
First cousins*	84	16.15	0.0625	0.010096154
Type I (Father's brother's daughter)	32	6.15		
Type II (Mother's sister's daughter)	17	3.27		
Type III (Father's sister's daughter)	12	2.31		
Type IV (Mother's brother's daughter)	23	4.42		
First cousins once removed	15	2.88	0.0313	0.000902885
Second cousins	16	3.08	0.0156	0.00048
Distant cousins	32	6.15	< 0.0156	
Total coefficient of inbreeding**				0.011719423

*For the degree of consanguinity "First Cousins," the type of mating is given from the male perspective

**Inbreeding coefficient up to 2nd cousins

Table 2 Associations between socio-demographic factors and consanguinity in the Souss population (N = 520)

Factors	Consanguineous*	Non- consanguineous*	Total	p-value
Women				
Age at first marriage				0.011
≤ 16	31.9	68.1	119	
17–22	33.5	66.5	227	
23–28	21.7	78.3	120	
29 +	14.8	85.2	54	
Mean age at first marriage (mean ± standard error)	19.84 ± 0.380	21.34 ± 0.317	20.91 ± 0.253	< 0.001
Birth environment (Rural/Urban)				0.914
Rural	28.5	71.5	491	
Urban	27.6	72.4	29	
Education level				0.308
Illiterate	28.3	71.7	421	
Quranic Pre-School	40.0	60.0	10	
Primary	33.3	66.7	51	
Secondary	30.4	69.6	23	
Higher	6.7	93.3	15	
Men				
Age at first marriage				< 0.001
≤ 16	0.0	100.0	4	
17–22	29.3	70.7	92	
23–28	38.4	61.6	190	
29 +	20.5	79.5	234	
Mean age at first marriage (mean ± standard error)	27.16 ± 0.514	29.81 ± 0.487	29.06 ± 0.381	< 0.001
Birth environment (Rural/Urban)				0.796
Rural	28.6	71.4	497	
Urban	26.1	73.9	23	
Education level				< 0.001
Illiterate	17.8	82.2	180	
Quranic pre-school	35.0	65.0	120	
Primary	41.0	59.0	134	
Secondary	21.6	78.4	51	
Higher	22.9	77.1	35	
Occupation status				< 0.001
Agriculture	19.3	80.7	109	
Commerce	41.8	58.2	165	
Labor/manual job	23.1	76.9	147	
Office job/services	24.2	75.8	99	
Couples with the same village/town of birth				< 0.001
Yes	45.0	55.0	218	
No	16.6	83.4	302	
Couples with the same tribe of birth				< 0.001
Yes	34.6	65.4	361	
No	14.5	85.5	159	
Parental consanguinity (n = 520)				< 0.001
Yes	43.9	56.1	107	
No	24.5	75.5	413	
Marriage cohort				0.602
< 1975	24.6	75.4	122	
1975–1989	31.6	68.4	177	
1990–2002	28.8	71.2	111	
2003–2019	27.3	72.7	110	

Table 2 (continued)

*Except for mean age at first marriage, the numbers are expressed in percentages (%)

Men's professional status is highly associated with consanguinity ($p < 0.001$), with a high prevalence of commercial activity among consanguineous couples.

Geographical endogamy based on birthplace, both at the tribal and village level, was significantly associated with consanguinity ($p < 0.001$). The involvement in consanguineous unions was found to be significantly correlated with parental consanguinity ($p < 0.001$). In other words, participants born of consanguineous unions tend to reproduce the same pattern as their parents.

The evolution of consanguinity, based on marriage contraction year, was not significant ($p > 0.05$), which confirms the attachment of the descendants to their parental matrimonial behavior.

The results of the multivariate logistic regression analysis are summarized in Table 3. The age at first marriage was found to be a significant predictor of consanguinity for both genders. For men, marrying between 23 and 28 increased the likelihood of consanguineous marriage by 1.99 times more than marrying at 29 and above (OR = 1.993; [95% CI 1.193–3.329]). Similarly, women who marry below the age of 23 have a significantly higher probability of being involved in consanguineous unions, with odds ratios of 3.478 (95% CI 1.255–9.636) and 2.863 (95% CI 1.127–7.270).

Based on our findings, the husband's professional status had a significantly higher impact on consanguinity than his educational level. Indeed, men who work in commercial fields are more likely to marry within their family (OR = 2.250; [95% CI 1.170–4.327]).

As regards the couples' birthplace (geographical endogamy), village endogamy appears to be a more significant matrimonial determinant than tribal endogamy. Indeed, couples with the same village/town of birth are 4.96 times more likely to be engaged in consanguineous marriages (OR = 4.960 [95% CI 2.824–8.713]). In addition, the likelihood of consanguinity among couples is 2.47 times higher when their parents are consanguineous (OR = 2.460 [95% CI 1.464–4.135]).

Table 3 Multivariate logistic regression analysis of socio-demographic factors associated with consanguinity in the Souss population

Socio-demographic characteristics	*p*-value	[95% CI]	Odds ratio
Age at first marriage (women)			
≤ 16	0.017	1.255–9.636	3.478
17–22	0.027	1.127–7.270	2.863
23–28	0.218	0.694–4.936	1.851
29 + *(Ref.)*			1.000
Age at first marriage (men)			
≤ 16	–	–	–
17–22	0.536	0.619–2.513	1.247
23–28	0.008	1.193–3.329	1.993
29 + *(Ref.)*			1.000
Men's education			
Illiterate	0.102	0.140–1.194	0.408
Quranic pre-school	0.638	0.461–3.536	1.277
Primary	0.531	0.499–3.858	1.387
Secondary	0.610	0.229–2.376	0.737
Higher *(Ref.)*			1.000
Men's occupation			
Agriculture	0.641	0.543–2.696	1.210
Commerce	0.015	1.170–4.327	2.250
Labor/manual job	0.915	0.517–2.088	1.039
Office job/services *(Ref.)*			1.000
Couples with the same village/town of birth			
Yes	< 0.001	2.824–8.713	4.960
No *(Ref.)*			1.000
Couples with the same tribe of birth			
Yes	0.765	0.462–1.764	0.903
No *(Ref.)*			1.000
Parental consanguinity ($n = 520$)			
Yes	0.001	1.464–4.135	2.460
No *(Ref.)*			1.000

Ref: Reference category

Biological effects of consanguinity on offspring health

To assess the impact of consanguinity on offspring health, we excluded 46 couples from our study. Specifically, we omitted couples without children, either because their marriages were recent compared to the survey period or because they involved women of advanced age.

Table 4 displays the inbreeding coefficients as well as the rates of different disease categories in the offspring of the studied couples. According to our findings, the inbreeding coefficient differed significantly between the three disease groups. The highest coefficient is observed in the category of congenital disabilities (0.0330538), followed by complex diseases (0.0252805), and infectious diseases (0.0072933). Similarly, diseased consanguineous offspring decreased from 67.31% for congenital disabilities to 47.15% and 20% for complex and infectious diseases, respectively.

The prevalence of common diseases among the offspring of consanguineous and non-consanguineous

Table 4 Rates and inbreeding coefficients of different disease categories in offspring

Categories	Total	Non-consanguineous offspring		Consanguineous offspring		Mean inbreeding coefficient
		n	%	*n*	%	
Ill offspring [a]	183	105	57.38	78	42.62	0.0242204
Types of diseases						
Congenital disabilities	52	17	32.69	35	67.31	0.0330538
Non-communicable diseases	123	65	52.85	58	47.15	0.0252805
Infectious diseases	30	24	80	6	20	0.0072933

[a] Individuals who have multiple types of diseases at the same time are counted only once in the total number of ill individuals

couples is presented in Table 5. The results show that offspring of consanguineous unions have a significantly higher risk of congenital disabilities than their non-consanguineous counterparts. This applies particularly to mental retardation (OR = 5.219; [95% CI 1.545–17.631]; $p = 0.008$), physical disability (OR = 3.753; [95% CI 1.398–10.074]; $p = 0.009$), and deafness-mutism (OR = 4.262; [95% CI 1.004–18.089]; $p = 0.049$). For complex diseases, the risk of cardiovascular disorders was significantly higher in the offspring of consanguineous than non-consanguineous couples (OR = 2.167; [95% CI 1.036–4.530]; $p = 0.040$). Other complex diseases (diabetes, cancer, bronchial asthma, epilepsy and psychiatric disorders) were more common in non-consanguineous marriages.

Discussion

Prevalence of consanguinity in the Souss region

Consanguinity in the Souss population was estimated at 28.46%, which was significantly higher than the reported national rate (23.4%) [24]. However, the prevalence of consanguineous unions in Morocco differs across regions. Higher levels were reported in the Middle Atlas and Tiflet areas, reaching 30.32% and 38.90%, respectively [25, 26]. Other studies conducted in Chaouia (25.38%), Tetouan (23.7%), and Doukkala (26.56%) revealed lower levels of consanguinity compared to our region [16, 27, 28].

Other studies in North African and Asian Muslim populations, such as Algeria, Tunisia, Egypt, Jordan, Oman and Pakistan [29–34] revealed higher prevalence of consanguinity compared to our region. In fact, the perpetuation of consanguinity at such frequencies could be justified by its social, cultural, and economic benefits. In Arab-Muslim societies, consanguineous marriages maintain family structure, stability, and property preservation [7, 35]. Other benefits include reduced marriage costs and dowry payments [7, 35].

The mean inbreeding coefficient reported in our study is 0.0117, with a preference for first-cousin unions, particularly the patrilateral parallel type (the father's brother's daughter). The same pattern was reported in other Muslim communities in North Africa [27, 29, 35] and Asia [5, 31, 36]. This preference could be justified

Table 5 Prevalence of common diseases among offspring of consanguineous and non-consanguineous couples

Diseases among offspring	Consanguineous *n* = 136 (%)	Non-consanguineous *n* = 338 (%)	OR	[95% CI]	*p*-value
Congenital disabilities					
Mental retardation	8 (5,9)	4 (1,2)	5,219	1,545–17,631	0,008
Deaf-mutism	5 (3,7)	3 (0,9)	4,262	1,004–18,089	0,049
Blindness	1 (0.7)	0	–	–	–
Physical disability	10 (7,4)	7 (2,1)	3,753	1,398–10,074	0,009
Complex diseases					
Cardiovascular diseases	14 (10,3)	17 (5,0)	2,167	1,036–4,530	0,040
Diabetes	11 (8,1)	16 (4,7)	1,771	0,800–3,922	0,159
Cancer	1 (0,7)	3 (0,9)	0,827	0,085–8,022	0,87
Bronchial asthma	4 (2,9)	6 (1,8)	1,677	0,466–6,038	0,429
Epilepsy	3 (2,2)	4 (1,2)	1,883	0,416–8,529	0,411
Psychiatric disorders	7 (5,1)	11 (3,3)	1,613	0,612–4,252	0,334

by pre-Islamic Arab tradition and Quranic provisions allowing women to inherit wealth [7].

Socio-demographic determinants of consanguineous marriages

Consanguinity and age at first marriage

In our study, the proportion of women who marry before age 23 was significantly higher than that of their late-marrying counterparts. This observation supports previous studies finding a significant relationship between early age at marriage and consanguinity [16, 36, 37]. On the other hand, men have a significantly higher likelihood (1.99 times) of contracting consanguineous unions between the ages of 23 and 28. However, Moussouni's study showed that for this age bracket, individuals are more likely to choose an unrelated partner than a close cousin [38].

Indeed, convinced by the tolerance and flexibility of young brides, Moroccan families marry their daughters at a young age [39]. However, this behavior has a negative impact on women since they marry before reaching emotional maturity [40].

Consanguinity and birth environment (Rural/ Urban)

In this study, there was no statistically significant difference in the frequency of consanguineous marriages between urban and rural birthplaces. The same observation was reported by Kaplan et al., in Turkey [36]. However, previous studies revealed significantly higher rates of consanguinity among couples of rural origin [16, 33, 35, 37]. The main explanation for our findings was the predominance of rural origin among couples. This situation resulted from the rural exodus (from rural areas) fostered by urbanization and regional climate changes [41]. However, the cultural and social values of the birth environment are so embedded in the individuals' subconscious that they reproduce them through their behavior [8]. A striking example was the high rate of consanguinity reported among descendants of Pakistani immigrants in the UK [42]. Thus, in our region, the influence of the birth environment on consanguinity will become evident in the future generation, especially in the degree of attachment to parental traditions.

Consanguinity and husband's occupation status

In contrast to several studies reporting a significant relationship between agricultural occupations and consanguinity [11, 16, 33, 43], this study found a higher probability for men working in commerce to be involved in consanguineous unions. Another study in Pakistan reported high levels of consanguinity among office workers and drivers [44].

Our observations support the previous hypothesis of rural exodus. Indeed, migration caused people to adapt to a different way of life, from agricultural to commercial occupations, while keeping their original cultural values. For his part, Jean Waterpoory has described the familial cohesiveness characterizing rural Soussi migrants who trade in metropolitan cities [45]. This solidarity is reminiscent of village cohesion, driven by the need to maintain family assets and sources of collective income [46].

Consanguinity and education level

In North African and Muslim communities, education level was a significant determinant of consanguinity [27, 31, 35, 36]. Indeed, social, economic, and cultural characteristics of the rural areas have contributed to a gender disparity in education. Our population is a striking example, with women being the most illiterate, while a significant proportion of men have received a basic education (primary or Quranic), allowing them to learn the fundamentals for their future occupation as traders. These elementary levels, however, do not provide knowledge of consanguinity and its repercussions. On the other hand, studies in Yemen and Tunisia have reported a high rate of consanguinity associated with higher levels of men's education [47, 48]. These facts illustrate the influence of familial and cultural pressures on future spouse selection, regardless of individuals' level of awareness [49].

Consanguinity and geographical endogamy

In Arab-Muslim communities, individuals express a sense of loyalty to their tribe, village, or clan, which is an extension of their patrilineal kinship [7]. Our population supports this thesis, with a high level of endogamy and a significant association between consanguinity and both tribal and village endogamy. Similarly, results reported in the Chaouia region recorded a high 3.29-fold probability for endogamous couples to be involved in consanguineous unions [16]. Indeed, regional and ethnic stratifications were considered factors increasing consanguinity, especially in small communities [50]. In our study, this fact could explain the higher incidence of consanguinity associated with village than tribal endogamy.

Parental consanguinity

In our study, parental consanguinity was associated with a higher likelihood of consanguineous unions. Other studies in northern Morocco, Algeria, Pakistan, and Qatar have reported similar results [27, 29, 33, 51]. These studies attest to the intergenerational transmission of consanguinity. Indeed, parents who are convinced by the success of their consanguineous marriage may encourage their offspring to reproduce the same pattern. Other studies have attributed the inheritance of this behavior to

the offspring's loyalty to the cultural and familial values in which they were brought up [35, 51].

Evolution of consanguinity

To study the evolution of consanguinity in the Souss region, we considered periods referring to the country's demographic and economic history. The consanguinity levels recorded in this study do not differ significantly between marriage years, albeit decreasing after 1975. However, the practice of consanguinity in the Chaouia region, which experienced the same historical events, showed a significant decrease [16]. These results confirm our previous observations on environmental and familial influences. Indeed, attachment to parental and social values strongly impacts the level and evolution of consanguinity.

Biological effects of consanguinity on offspring health

Consanguinity and congenital disabilities

Numerous epidemiological studies have demonstrated the impact of consanguinity on children's health, particularly highly consanguineous communities [5]. Our study found a statistically significant difference in congenital disabilities based on consanguinity status, including mental retardation, physical disabilities, and deafness. Unfortunately, due to a lack of cases, we could not obtain information on blindness. However, other studies reported a significant association between consanguinity and both congenital and childhood blindness [52–54].

We recorded a significant risk of mental retardation in consanguineous offspring. Similar findings were reported in Tiflet (Morocco), Algeria, and Lebanon [25, 52, 55]. However, according to a Moroccan national study, the rate of consanguinity among patients with Down syndrome, which is one of the most common examples of gonadal non-disjunction, is relatively low at 15.25% [56]. On the other hand, previous studies have reported a high prevalence of intellectual disability and cognitive impairment in the progeny of consanguineous unions [10, 57].

Physical disabilities were the most common birth defects in consanguineous offspring (7.4%), with a 3.75-fold higher risk than in their non-consanguineous counterparts. Our results are consistent with two reported studies from Bangladesh and Lebanon, where consanguinity was associated with a high frequency of congenital anomalies [11, 52].

Furthermore, we found a statistically significant difference in sensory impairment, namely deafness-mutism, based on consanguinity status. Similarly, a Tunisian study reported a tenfold higher risk of deafness among consanguineous offspring than in their non-consanguineous counterparts [58].

Consanguinity and complex diseases

Of all the complex diseases studied, consanguinity was found to be a significant predictor only for cardiovascular conditions. No significant association was found between consanguinity and other diseases, including diabetes, cancer, bronchial asthma, epilepsy, and psychiatric disorders. However, in some studies conducted in Morocco, Qatar and Croatia, consanguinity was found to be associated with a wide range of non-communicable diseases, including epilepsy, asthma, leukemia, diabetes mellitus, cardiovascular disease, stroke, cancer, and uni/bipolar depression [12, 51, 59].

Our population displays a specific reproductive pattern, with a high prevalence of both consanguinity and endogamy. Through the cumulative effect of consanguinity, this pattern enriches the population gene pool with founder mutations inherited from a common ancestor [60]. Thus, the number of mutation carriers increases in extended families, resulting in high homozygosity [61]. This situation allows the appearance of recessive genetic disorders, including mental and physical disabilities [62].

Multiple studies analyzing genetic profiles of Moroccan populations have provided evidence supporting the previous conclusions. These studies have revealed an excess of homozygosity, which disrupts the genetic equilibrium. The authors attributed this phenomenon primarily to the high prevalence of consanguinity within these populations [63–65].

The environmental component contributes significantly to the incidence of non-communicable diseases, particularly complex and late-onset diseases. Indeed, the greater the impact of environmental factors, the more diluted the effect of consanguinity on disease incidence [66].

Our observations support this conclusion, revealing a noteworthy decrease in consanguinity coefficients as the likelihood of etiological environmental factors increases—shifting from congenital disabilities to infectious diseases. This suggests a diminished role of the genetic component, specifically in terms of homozygosity, in the disease's etiology. Our results highlight this trend, as non-consanguineous offspring demonstrate higher rates of non-communicable and infectious diseases, whereas consanguineous offspring predominantly manifest congenital disabilities.

In addition to non-genetic factors, the implication of common disease alleles in the gene pool, according to the common disease/common variant hypothesis, may reduce the influence of consanguinity on complex disorders [3].

In highly endogamous populations, the genetic intermingling between subpopulations (communities) is limited, resulting in intra-community genetic homogeneity

[67]. This leads to the accumulation of homozygosity regardless of consanguinity status, in other words an increase in random consanguinity [3]. From an epidemiological and clinical perspective, population stratification can mask the health effects of consanguinity. In other words, endogamy causes an underestimation of morbidity and mortality associated with consanguinity [30].

Limitations

This study highlights the impact of consanguinity on congenital and complex diseases in the Souss region. However, there are several limitations. First, given the cross-sectional nature of the study, the causal relationship between consanguinity and disease was not clearly established. Furthermore, individuals are likely to have a recall bias when it comes to information about their couple, parents, or children. However, the participation of families in the collective interview, as well as the availability of documents due to our presence in the family's home, allowed us to reduce the recall bias. Additionally, the limited number of individuals with certain reported conditions may, in statistical terms, affect the generalizability of the findings.

Finally, the ancestral data are limited to participants' parents, particularly in relation to parental consanguinity (partners' parent's data are not included). It should also be noted that there may be a bias regarding later-onset diseases, which can occur later in life for any individual (consanguineous and non-consanguineous), including cancers, cardiovascular diseases and diabetes. We have, however, made efforts to mitigate this bias to some extent by incorporating familial accounts of medical histories and ages of disease onset, taking advantage of our presence within the family household.

Conclusions

Based on the results presented and discussed above, we concluded that the population of Souss was not an exception to the Arab-Muslim communities in terms of high prevalence of consanguinity. This behavior was associated with socioeconomic and demographic factors, including early age of marriage, low level of education and endogamy.

This study highlighted the negative impact of consanguinity on health, including a high incidence of mental disorders, physical disabilities, deafness-mutism, and cardiovascular diseases. However, despite this fact, the population of the Souss region cannot be prevented from contracting such unions. On the one hand, parental influence on their offspring's marital choice contributes to the continuation of this behavior; on the other hand, the socioeconomic benefits provided by this type of marriage, mainly financial, emotional, and familial stability, encourage recent generations to opt for consanguineous unions.

In such a population scenario, the only possible interventions may be at two levels: awareness of the consequences of consanguinity and genetic counseling for related couples. Genetic screening tests will allow for the early detection of genetic disorders or malformations. Subsequently, this will provide early care for consanguineous children, enhancing, at the very least, their life expectancy and quality of life [5].

We are unable to make conclusions about possible solutions to this problem given to the cross-sectional design of this study. Our objective was to draw attention to the level of consanguinity in the Souss population and its impact on offspring health. In order to establish a clear causal relationship between consanguinity and disease, a genetic study based on an epidemiological survey would be necessary. To enhance the precision of our analysis, it is recommended to employ more specific methods, such as calculating the inbreeding coefficients derived from Runs of Homozygosity, commonly known as Froh. This method precisely quantifies both the proportion and length of homozygous segments in the genome. Utilizing such methods is essential to reduce the bias caused by random consanguinity in the stratified population [3].

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Author contributions

AH, HE and JT have contributed to the conception and design of the study, correction of the manuscript, and general supervision. KC, AE, and LH have participated in the fieldwork, data analysis, and interpretation. ND has contributed to the conception of the study, fieldwork, analysis and interpretation of data, and final drafting of the submitted manuscript.

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Availability of data and materials

The data used to support the conclusions of this work are available upon request from the corresponding author.

Declarations

Ethics approval and consent to participate

Ethical approval for this study was obtained from the Biomedical Research Ethics Committee (CERBC) of the Faculty of Medicine and Pharmacy of Casablanca, in accordance with the standards of the Declaration of Helsinki (v.2008).

Consent for publication

Not applicable.

Competing interests

The authors have no Competing interests to declare regarding the publication of this article.

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