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The prevalence of orofacial clefts in Qatar : a cross-sectional nationwide study

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Abstract

Background Cleft lip and palate are congenital craniofacial anomalies that significantly impact individuals and their families, both medically and psychosocially. The Qatari population has unique characteristics that are suggestive of a high prevalence of congenital anomalies: high consanguinity rate, large family size, advanced paternal age and high prevalence of certain genetic disorders. The lack of existing data concerning the epidemiology of cleft lip and/or palate in Qatar warrants a descriptive study addressing this topic.

Methods A retrospective nationwide hospital-based cross-sectional study conducted to determine the prevalence of orofacial clefts in Qatar from 2016 to 2021. Data were extracted from the corporation's Cerner database and electronic health records. Information concerning the cleft's characteristics, type, affected side, patient's gender, presence of associated syndromes, nationality, and maternal age were collected.

Results Out of the 147,727 live births, 148 had an orofacial cleft. The prevalence of cleft lip and/or palate was determined to be 1 per 1000 livebirths (95% CI: 0.85, 1.18). The prevalence of cleft lip was 0.18 (95% CI: 0.12, 0.27), cleft palate 0.39 (95% CI: 0.30, 0.51), and cleft lip and palate 0.43 (95% CI: 0.33, 0.55). Qataris had a prevalence of CL 0.25, CP 0.40, and CLP 0.56, compared to 0.16, 0.39, 0.39 for non-Qataris (*p*-value 0.186). Unilateral clefts predominated over bilateral (74.4% and 25.6%, respectively). Among the unilateral cases, 70.2% occurred on the left side. Most cases were isolated clefts, with only 10.2% having associated syndromes.

Conclusions The prevalence of orofacial clefts in Qatar is consistent with the globally reported prevalence. Most cases were unilateral and on the left side. Associated syndromes were infrequent and more common with cleft palate alone. Intriguing patterns were revealed between Qatari nationals and non-Qatari residents, with specific subtypes of orofacial clefts showing higher prevalence among nationals.

Keywords Cleft lip, Cleft palate, Cleft lip and palate, Prevalence, Electronic Health Records

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Introduction

Orofacial clefts (OFCs) describe a range of neonatal anomalies affecting the lips and the oral cavity, which include cleft lip, cleft lip and palate, and cleft palate alone [1]. OFCs are the most common craniofacial anomalies and one of the most common congenital deformities [2], occurring in 1 per 500–1000 births globally, with variation across geographic areas and ethnic groups [3]. A child with OFC suffers substantial morbidity including facial deformity, speech anomalies, feeding difficulties, recurrent ear infections and hearing loss [2]. Therefore, the condition poses significant medical, psychological, social, and financial implications on the affected individuals and families, with disparity within and between countries.

OFCs have a complex etiology with both genetic and environmental contributions. Although genes play a significant role in congenital deformities, the environment plays a crucial role in modulating gene effects. Risk factors such as parental consanguinity, maternal smoking, alcohol consumption, and illness during pregnancy have all been linked to OFCs [4]. On the other hand, vitamins and zinc supplementations early during pregnancy have been associated with decreased risk, in a dose-response manner [5, 6].

Descriptive epidemiologic studies on cleft lip and/or palate have been carried out in many countries, showing differences in the geographical distribution of clefts across the world. The results of such epidemiological studies provide data to determine the magnitude of the problem, the efforts needed to improve the quality of life for affected patients and the effectiveness of interventions. The Qatari population has unique characteristics that are suggestive of a high prevalence of congenital anomalies: high consanguinity rate, large family size, advanced paternal age and high prevalence of certain genetic disorders [7]. However, to the best of our knowledge, no epidemiological study of OFCs has been performed in Qatar. The lack of existing data concerning the epidemiology of cleft lip and/or palate in Qatar warrants a descriptive study addressing this topic. Consequently, this research aims to investigate the prevalence and associated features of OFCs in Qatar. The specific objectives of this study encompass estimating prevalence rates, analyzing the geographical and socio-demographic distribution, and investigating distinctive characteristics related to cleft lip and/or palate within the Qatari population.

Materials and methods

A quantitative descriptive nationwide hospital-based cross-sectional study was conducted to ascertain the prevalence of cleft lip and/or palate. This retrospective investigation encompassed all live births delivered at Hamad Medical Corporation in Qatar over a 6-year period, spanning from January 1, 2016, to December 31, 2021.

Hamad Medical Corporation (HMC) serves as Qatar's primary public healthcare provider, overseeing numerous hospitals, emergency services, specialized care centers, ambulatory facilities, as well as the national ambulance and home healthcare services. HMC hospitals, which feature dedicated maternity departments to provide specialized care for pregnant women and facilitate the childbirth process, operate across the country with a network of four medical campuses strategically positioned in distinct locations. These campuses cover the capital and extend to the North, South, and West regions of the nation. Therefore, we believe that the data derived from this hospital-based study effectively mirror those of a national based study. In fact, a comparison between the total number of live births at HMC and the national live birth figures of the same period, obtained from the Qatari National Census and Statistics Department, revealed that approximately 89% of all live births in Qatar during the study period occurred at HMC (Appendix Table 1).

Data were extracted from the corporation's Cerner database, capturing the total number of newborns and the number of babies born with cleft lip and/or palate between January 1, 2016, and December 31, 2021. Stillbirths and aborted fetuses were excluded from the study. After obtaining Institutional Review Board (IRB) approval from Hamad Medical Corporation (MRC-01-22-743), access was granted to electronic medical records pertaining to cleft cases and their respective mothers, facilitating the retrieval of information concerning the cleft's characteristics (cleft lip, cleft palate, cleft lip and palate), type (unilateral, bilateral), affected side (right, left), patient's gender, presence of associated syndromes, nationality, and maternal age. Informed consent was waived by the IRB because it is a minimal risk retrospective chart review in which no patient interaction will occur.

Data analyses was performed using Stata/SE ™ statistical software (version 11.1). Descriptive statistics were calculated for the cleft prevalence per 1000 total live births. Mann-Whitney as well as Fishers' exact tests were performed to test for statistical significance. The Clopper–Pearson interval (exact method) was used for calculating binomial confidence intervals of the prevalence measures. All statistical tests were two-tailed and *P*-values less than 0.05 were considered statistically significant.

Results

The study encompassed a sample of 147,727 live births that occurred at Hamad Medical Corporation between the years 2016 and 2021, constituting approximately 89% of all live births in Qatar within the same time period.

Among these births, 148 cases (0.1% of total live births) were diagnosed with orofacial clefts, comprising 84 male and 64 female. The average maternal age among cases was 30.5±5.5 years, with fairly uniform distribution across the different birth seasons of the year (Table 1).

Out of the 147,727 live births, 22% were Qatari nationals, while the remaining 78% represented residents of various other nationalities in Qatar. The incidence of orofacial clefts was 0.12% in Qatari nationals and 0.09% in non-Qatari residents. Despite a slightly elevated percentage of males among non-Qataris (58.7%) in contrast to Qataris (51.3%), this gender-based difference did not achieve statistical significance (p-value 0.455). Notably, no statistically significant distinctions emerged between Qatari and non-Qatari populations across multiple assessed variables, including the presence of orofacial clefts, birth year, gender distribution, maternal age, birth season, cleft nature, cleft type, and cleft side (Table 1). Similarly, there were no significant variations in these assessed variables when comparing males and females (Appendix Table 2).

Throughout the 6-year study period, the overall prevalence of cleft lip and/or palate was determined to be 1 per 1000 live births (95% confidence interval: 0.85, 1.18). Among these, the prevalence of isolated cleft lip was observed at the lowest rate of 0.18 per 1000 (95% CI: 0.12, 0.27), while the prevalence of cleft palate alone along with cleft lip and palate was comparable at 0.39 (95% CI: 0.30, 0.51) and 0.43 per 1000 (95% CI: 0.33, 0.55), respectively (Table 2). Qataris had a slightly increased prevalence of orofacial clefts at 1.21 per 1000, compared to the 0.94 per 1000 for non-Qataris. They exhibited an increased prevalence of cleft lip with or without a palate and a similar prevalence of cleft palate alone (Table 2). However, the

Table 1 Descriptive statistics for Socio-demographic and cleft characteristics among Qatari Nationals and Residents

Variables	Qataris *N* (%)	Residents *N* (%)	Total *N* (%)	*P*-value*
		Total live birth		
Presence of orofacial cleft				
Yes	39 (0.12%)	109 (0.09%)	148 (0.10%)	0.196
No	32,258 (99.92%)	115,321 (99.94%)	147,579 (99.93%)	
		Cleft patient demographics		
Year of birth				
2016	7 (18.0%)	19 (17.4%)	26 (17.5%)	0.618
2017	8 (20.5%)	23 (21.1%)	31 (20.1%)	
2018	9 (23.1%)	12 (11.0%)	21 (14.2%)	
2019	4 (10.3%)	14 (12.8%)	18 (12.2%)	
2020	5 (12.8%)	19 (17.4%)	24 (16.2%)	
2021	6 (15.4%)	22 (20.2%)	28 (18.9%)	
Gender				
Males	20 (51.3%)	64 (58.7%)	84 (56.8%)	0.455
Females	19 (48.7%)	45 (41.3%)	64 (43.2%)	
Mother's age at birth				
Number	39	109	148	0.4663
Mean (SD)	30.0 (±5.4)	30.7 (±5.2)	30.5 (±5.3)	
Season of birth				
Spring	13 (33.3%)	21 (19.3%)	34 (23.0%)	0.315
Summer	9 (23.1%)	37 (33.9%)	46 (31.1%)	
Fall	8 (20.5%)	26 (23.9%)	34 (23.0%)	
Winter	9 (23.1%)	25 (22.9%)	34 (23.0%)	
		Cleft characteristics		
Cleft nature				
Cleft lip	8 (20.5%)	19 (17.4%)	27 (18.2%)	0.709
Cleft Palate	13 (33.3%)	45 (41.3%)	58 (39.2%)	
Cleft lip and palate	18 (46.2%)	45 (41.3%)	63 (42.6%)	
Cleft lip with or without palate type				
Unilateral	21 (80.8%)	46 (71.9%)	67 (74.4%)	0.436
Bilateral	5 (19.2%)	18 (28.1%)	23 (25.6%)	
Cleft lip with or without palate side				
Left	15 (71.4%)	32 (69.6%)	47 (70.2%)	0.543
Right	6 (28.6%)	14 (30.4%)	20 (29.9%)	

*P-values reported for Fisher's exact tests for categorical variables and Mann-Whitney U test for continuous and ordinal variables

Table 2 The numbers and prevalence of orofacial clefts among Qatari Nationals and Residents

Cleft Type	Qataris		Residents		Total	
	N of cases	Pr (95% CI)	*N* of cases	Pr (95% CI)	*N* of cases	Pr (95% CI)
Cleft Lip	8	0.25 (0.11, 0.49)	19	0.16 (0.10, 0.26)	27	0.18 (0.12, 0.27)
Cleft Palate	13	0.40 (0.21, 0.69)	45	0.39 (0.28, 0.52)	58	0.39 (0.30, 0.51)
Cleft Lip and Palate	18	0.56 (0.33, 0.88)	45	0.39 (0.28, 0.52)	63	0.43 (0.33, 0.55)
Cleft Lip and/or Palate	39	1.21 (0.86, 1.65)	109	0.94 (0.78, 1.14)	148	1.00 (0.85, 1.18)

N: number of cases, Pr: prevalence per 1000 live birth, CI: confidence interval

difference was not statistically significant (p-value 0.186). Within the Qatari population, the prevalence of orofacial clefts exhibited minor fluctuations over the study duration, with an increase observed in 2018 (Appendix Table 3).

Unilateral cleft lip with or without palate predominated over bilateral clefts, accounting for 74.4% and 25.6% of cases, respectively. Additionally, the majority of the unilateral clefts occurred on the left side among both Qataris (71.4%) and non-Qataris (69.6%), constituting 70.2% of all unilateral cleft cases. Occurrences of associated syndromes were infrequent, with only 15 cases (10.2%) displaying cleft lip, cleft palate, or both, in conjunction with various syndromes. The most common syndrome associated was Pierre Robin sequence with 6 out of the 15, other syndromes included were: ectrodactyly ectodermal dysplasia, Down Syndrome, DiGeorge syndrome, CHARGE syndrome, Microdeletion syndrome, Orofaciodigital syndrome and Edwards syndrome. Among the 15 syndromic cases, 12 were associated with cleft palate alone, and 3 were associated with cleft lip and palate.

Discussion

Cleft lip and palate are congenital craniofacial anomalies that significantly impact individuals and their families, both medically and psychosocially. This prevalence study on orofacial clefts in Qatar holds unique significance as the first of its kind within the country. It encompasses a substantial sample representing around 89% of all live births registered in Qatar between 2016 and 2021. It provides valuable insights into the prevalence, demographic patterns, and characteristics of orofacial clefts in Qatar.

The observed overall prevalence of cleft lip and/or palate in Qatar was 1 per 1000 live births. Among the study population, Qatari nationals exhibited a slightly higher prevalence (1.2/1000) compared to Qatari residents (0.9/1000). Both figures are in line with the latest reported estimates of the global prevalence of orofacial clefts, which is 1.1 per 1000 live births. Highest reported prevalence of cleft lip and/or palate been reported for Native Americans (3.6/1000) and Asians (1.82/1000), while Africans had the lowest prevalence rates (0.3/1000) [8–10].

Comparing the prevalence of orofacial clefts in Qatar to other Gulf countries, we found a few studies carried out in Saudi Arabia, UAE, and Oman [11–16]. The overall prevalence of orofacial clefts reported in these studies varied greatly. In Saudi Arabia, for instance, the reported prevalence ranged from 0.3 to 2.19 per 1000 live births [11, 12]. This variation was attributed to the absence of a universal registration system [13].

Cleft lip alone had the lowest prevalence in our study (0.18/1000), which was lower than both cleft palate alone (0.39/1000) and cleft lip and palate (0.43/1000). A recently published meta-analysis in 2021 reported a higher global prevalence of cleft lip (0.33/1000) compared to our sample [3]. Nevertheless, this meta-analysis included numerous studies reporting similar prevalence rates to ours. In fact, the prevalence of cleft lip ranged from as low as 0.06 up to 0.85 [8, 17]. On other hand, the reported global prevalence of cleft lip and palate as well as cleft palate alone (0.45 and 0.33, respectively) are more comparable to the prevalence found in our sample.

Our study uncovered intriguing patterns in orofacial cleft prevalence between Qatari nationals and non-Qatari residents. While the overall prevalence was slightly higher among Qataris, specific subtypes showed notable variations. In particular, Qatari nationals exhibited a higher prevalence of cleft lip alone (0.25 compared to 0.16) and cleft lip with palate (0.56 compared to 0.39). However, the prevalence of cleft palate alone remained consistent (0.39 and 0.40). It should be highlighted that the observed differences were not statistically significant; this could be due to the rarity of orofacial clefts, leading to small differences that might make it challenging to establish conclusive statistical significance.

Similarly, the higher prevalence of orofacial clefts observed among Qatari males (1.1/1000) compared to females (0.9/1000), did not reach statistical significance. The difference was particularly pronounced for the prevalence of cleft lip in males (0.24/1000), double the prevalence in females (0.12/1000), which aligns with findings from previous studies indicating a higher prevalence of OFCs among males [18, 19].

In this study, the majority of cleft lip with or without palate cases were unilateral (74%), with more than two-thirds of them occurring on the left side. This observation aligns with several previously published studies [11, 14, 20]. While the mechanism behind this prevalence on the left side remains unclear, one suggested reason is the

slower development of the facial artery on the left side compared to the right [21].

As anticipated, the majority of orofacial clefts presented as isolated defects. Occurrences of associated syndromes were infrequent, with only 10% of cases were displaying concurrent orofacial clefts with various syndromes. Among the syndromic cases, 80% were associated with cleft palate alone, and 20% were associated with cleft lip and palate. This finding supports existing reports, which suggests that cleft palate alone tends to be more commonly associated with syndromes compared to cleft lip with or without palate [18, 22].

While the case information was obtained from medical records, the level of clinical detail available for this analysis is limited. There was variation in the recorded covariates, with limited information on consanguinity as well as family history of orofacial clefting. This limitation hindered our ability to thoroughly assess the significance of different risk factors within the Qatari population. In the future, the establishment of a standardized register system with more comprehensive data related to patients with orofacial clefts in HMC (Hamad Medical Corporation) is essential. Such a database will aim to gather birth, demographic, and epidemiological data of all children born in Qatar with a cleft lip and/or palate, alongside compiling information regarding the treatments administered to these children and the subsequent outcomes of such interventions.

In conclusion, this study disclosed an overall prevalence rate of orofacial clefts in Qatar at 1 per 1000 live births with higher prevalence among Qatari nationals compared to residents. Gender-based variations were also observed, with males showing a higher prevalence, particularly for cleft lip. However, these observed differences did not achieve statistical significance. Additionally, the majority of cases were unilateral, with more than two-thirds of them occurring on the left side. Associated syndromes were relatively infrequent and more common in cleft palate alone cases. These findings provide valuable insights into the epidemiology of orofacial clefts in the Qatari population.

Supplementary Information

The online version contains supplementary material available at https://doi.org/10.1186/s12903-024-04455-8.

Supplementary Material 1

Author contributions

M.K. contributed to conception and design, data acquisition, analysis, and interpretation, and drafted the manuscript. M.H. contributed to data acquisition and critically revised the manuscript. M.B contributed to data acquisition and critically revised the manuscript. N.H. contributed to conception and critically revised the manuscript. All authors gave their final approval and agreed to be accountable for all aspects of the work.

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Data availability

The datasets generated and/or analyzed during the current study are not publicly available due to the institution regulations, however, they are available from the corresponding author on reasonable request.

Declarations

Ethics approval and consent to participate

This study received approval from the Institutional Review Board (IRB) at Hamad Medical Corporation under reference number MRC-01-22-743. Informed consent was waived by the Hamad Medical corporation's IRB because it is a minimal risk retrospective chart review in which no patient interaction will occur.

Consent for publication

Not Applicable.

Competing interests

The authors declare no competing interests.

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