MOLECULAR EPIDEMIOLOGY OF ALPHA THALASSEMIA MUTATIONS IN DUHOK CITY, KURDISTAN REGION OF IRAQ: A 9-YEAR RETROSPECTIVE STUDY

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ABSTRACT

Background: Alpha thalassemia demonstrates high prevalence in Iraq, particularly among consanguineous Kurdish subgroups. However, data remains limited regarding the molecular epidemiology of causative alpha globin mutations in this population. This study aimed to elucidate the mutational spectrum and temporal distribution of alpha thalassemia alleles among native inhabitants of Duhok City in northern Iraq.

Methods: In this retrospective cross-sectional study, positive alpha globin genetic testing records of 1,295 Duhok natives at a specialized northern Iraqi laboratory from 2014-2022 were analyzed to determine mutant genotype and allele frequencies.

Results: The five most common mutant genotypes detected were -α3.7/-α3.7 (5.5%), -α3.7/αα (19.1%), -αMED/αα (3.2%), -α3.7/-αMED (2.6%) and ααIVS1-5nt/αα (1.5%). The -α3.7 deletion was predominant affecting 43.8% of alleles. A steady 9-year upward annual detection trajectory was observed.

Conclusions: Population-specific molecular analysis may provide seminal evidence to shape thalassemia screening and prevention policies targeting the high-risk marginalized Kurdish community in northern Iraq.

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Keywords: alpha thalassemia, mutations, molecular epidemiology, Duhok City, Iraq, Kurdistan.

Alpha thalassemia is among the most common monogenic disorders globally, reaching carrier frequencies as high as 80-90% in certain Asian regions [(1)]. It particularly affects tropical/subtropical countries spanning the Mediterranean region, Middle East, Indian subcontinent, Southeast Asia and Southern China[(2,3)]. Alpha thalassemia results from mutations in the HBA1 and HBA2 alpha globin genes located on chromosome 16, inherited in an autosomal recessive manner.[(4)] Over 100 disease-causing variants are described, exhibiting significant geographical heterogeneity[(5)]. The predominant α+ thalassemia alleles are deletions removing one (-α) or both (--) linked alpha genes, including -α3.7 (single gene deletion), -α4.2 (single gene deletion), and - -MED (double gene deletion), while α0 thalassemia arises from inactivation point mutations[(2)].

Imbalanced α/non-α globin chain production causes ineffective erythropoiesis and hypochromic microcytic anemia of varying severity based on the number of affected alleles[(1)]. Silent carrier states with one or two mutated alpha genes may only manifest minimal hematological changes. However, interaction of α0 and α+ alleles produces hemoglobin H disease, whereas homozygosity or compound heterozygosity for α0 mutations is incompatible with life causing hydrops fetalis[(2)]. Alpha thalassemia represents a significant public health issue throughout the Mediterranean, Middle East and Indian

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subcontinent regions[2]. Approximations indicate over 5% of the global population are carriers, with 60,000 severely affected annual births, majority concentrated in low and middle income countries[6].

Iraq constitutes a thalassemia high-prevalence zone with reported carrier rates between 25-30% in the general population[7]. The heterogeneous Iraqi society includes distinct ethnic fractions like Arabs and Kurds with high consanguinity perpetuating autosomal recessive conditions[8-10]. Kurds reside across mountainous northern Iraq, representing 15-20% of the nation's populace[11]. They harbor unique yet overlapping profile of hemoglobinopathies compared to ethnic Arabs[12]. Neighboring Iran and Turkey also demonstrate elevated alpha globin mutation frequencies[13-15]. However, Iraqi Kurds tend to intermarry within isolated tribal clans promoting recessive genetic disorders[7,16,17]. The province of Duhok borders Syria and Turkey at the northern frontier of Iraqi Kurdistan with probable distinctive alpha thalassemia mutation repertoire[18,19].

Duhok City is located in the Duhok governorate of Iraqi Kurdistan near the borders with Turkey and Syria. It has an estimated population of over 1,470,000 inhabitants, the majority being ethnic Kurds.[20] This region has historically experienced high rates of tribalism and consanguineous marriage. It is geographically isolated from central and southern parts of Iraq. These factors contribute to increased prevalence of recessive hereditary blood disorders among the native population[21,22].

Despite recognizably high prevalence across Iraq, the distribution of particular alpha globin mutations among various ethnic and regional subgroups is less comprehensively characterized[23]. Available reports catalogue common Middle Eastern variants like -α3.7, - -MED and ααIVS-I, but information from Iraqi Kurdistan and specifically Duhok city is limited[3,24]. Duhok City is situated near the Syrian and Turkish borders, representing a geographically isolated region of the Iraqi Kurdistan territory. Its predominantly Kurdish tribal inhabitants exhibit high consanguinity and accordingly increased risk for autosomal recessive hemoglobinopathies like alpha thalassemia[22]. Defining the relative proportions of α thalassemia mutations within communities directed by molecular screening and diagnosis[25,26] and allow risk-stratification for genetic counselling plus enables targeted prenatal or preimplantation interventions to prevent disease in high-risk populations like the Kurds[27]. As northernmost Iraqi province, Duhok may demonstrate unique mutational patterns compared to central parts of the country The study aimed to characterize the molecular epidemiology of mutant alpha globin genes specifically among the native Duhok Kurds over a 9-year timeframe spanning 2014-2022 based on diagnostic laboratory records. The findings provide seminal evidence guiding clinical recommendations and public health policies for α thalassemia prevention in this marginalized population.

PATIENTS AND METHODS

Study Design and Setting

We performed a quantitative retrospective cross-sectional study through secondary analysis of archived alpha globin genetic testing data from patients originating in Duhok City, Iraq. These data were provided from genetic analysis laboratory in Amr Lab in Duhok city / Kurdish region of northern Iraq.

Inclusion and Exclusion Criteria

The sample comprised all consecutive cases between 2014-2022 with positive results for alpha thalassemia mutations

among native Duhok inhabitants assessed at AmrLab. patients of Iraqi origin from other provinces, those lacking confirmed Duhok residence and tests yielding negative or inconclusive outcomes have been excluded. These criteria ensured the cohort represented an ancestrally homogeneous ethnic Kurdish subgroup for site-specific analysis.

Variables and Data Sources

The anonymized dataset was extracted from Amr Lab records into a standardized form. It documented demographic characteristics namely age, gender, family history and details of detected alpha globin mutations including genotype and allele variants using standard human genome reference notation. Available peripheral blood hematological indices were also registered.

Alpha thalassemia diagnosis entailed multiplex gap-polymerase chain reaction (PCR) for 21 Middle Eastern alleles including common large deletions, small deletions and point mutations according to protocols by Tan et al[28]. This allowed efficient detection of regionally prevalent variants.

Sample Size

The analysis encompassed all eligible positive cases over the 9-year study timeframe totalling 1,295 patients. This number represented an adequate sample to ascertain statistically robust mutation frequencies and distribution patterns in the native Duhok Kurd population.

Ethical Considerations

Institutional ethics approval was obtained prior to accessing the anonymized dataset. Individual consent was exempted given retrospective analysis of recorded health information. The data were maintained confidentiality throughout the study. The molecular epidemiological findings aimed to guide clinical recommendations for thalassemia screening and diagnosis to promote the health of the indigenous Kurdish subgroup.

STATISTICAL ANALYSES

Descriptive statistics analysis illustrated the overall and genotype-specific mutant alpha globin allele frequencies using numbers and percentages. Stratified analysis examined demographic factors associated with particular globin mutations. Temporal annual trends were evaluated across the study duration. Duhok City patterns were compared to previous Iraqi and regional Kurdish thalassemia reports wherever possible.

RESULTS

The study sample constituted 1,295 Duhok native patients with confirmed alpha thalassemia mutations on genetic analysis over a 9-year period. The cohort was 57% male and 43% female ranging from infancy to 60 years, although majority were children or young adults undergoing routine screening. Positive family history was elicited in 74%. Table 1 shows the outlines of absolute genotype frequencies among the thalassemia cases. The five most regularly detected variants in order of descending prevalence were: -α3.7/-α3.7 (5.48%), -α3.7/αα (19.14%), - -MED/αα (3.17%), -α3.7/- -MED (2.63%), and αα2 IVS1-5nt/αα (1.47%). The homozygous -α3.7/-α3.7 genotype represented the most common mutant alpha globin diplotype among 5.48% cases, depicting interaction between two deleted alleles. The normal gene frequency in heterozygotes like -α3.7/αα (19.14%) indicated over one-third Duhok Kurd chromosomes harboured this deletion. Compound heterozygous genotypes with two different mutations such as -α3.7/- -MED (2.63%) can produce clinically severe hemoglobin H disease

Table 1: Alpha thalassemia genotype distribution among patients from Duhok City, Iraq (2014-2022)

Genotype	Frequency	Percentage
-3.7 heterozygous (-3.7/aa)	248	19.14%
-3.7 homozygous (-3.7/-3.7)	71	5.48%
--MED heterozygous (--MED/aa)	41	3.17%
-3.7/--MED compound heterozygous	34	2.63%
Heterozygous for a2 IVS1-5nt mutation	19	1.47%
Heterozygous for a2 poly A-2 mutation	14	1.08%
Heterozygous for a2 poly A-1 mutation	13	1.00%
-4.2 heterozygous (-4.2/aa)	12	0.93%
--20.5kb heterozygous (--20.5kb/aa)	5	0.39%
-3.7/-4.2 compound heterozygous	4	0.31%
--FIL heterozygous (--FIL/aa)	4	0.31%
Homozygous for a2 IVS1-5nt mutation	3	0.23%
Homozygosity of a2 poly A-2 mutation	3	0.23%
-4.2 homozygous (-4.2/-4.2)	2	0.15%
-3.7/-20.5 kb compound heterozygous	2	0.15%
--MED/a2 poly A-2 compound heterozygous	1	0.08%
--MED/a1 cd 59 [G>A] (Hb Adana) compound heterozygous	1	0.08%
Homozygosity of a2 poly A-1 mutation	1	0.08%
Homozygosity for a2 cd 19 [-G] mutation	1	0.08%
IVS1-5n homozygous	1	0.08%
anti-3.7 gene triplication (aaa/aa)	1	0.08%
Total	481	

Table 2 displays the corresponding allele-level mutation rates. Here -α3.7 constituted over 43% alleles followed by the normal gene (35.6%), while - -MED accounted for 6.3%. This breakdown delineated relative proportions of the common mutant alpha globin alleles circulating among Duhok natives. The predominance of -α3.7 agreed with its high representation across Middle East Arab subgroups, although the descending order of other regional variants somewhat diverged[10, 22].

Table 2: Alpha globin allele frequencies among Duhok City natives with thalassemia (2014-2022)

Allele	Frequency	Percentage
-3.7	567	43.8%
Normal alpha gene (aa)	461	35.6%
--MED	82	6.3%
a2 IVS1-5nt	38	2.9%
a2 poly A-2	28	2.2%
a2 poly A-1	26	2.0%
-4.2	24	1.9%
--20.5kb	10	0.8%
--FIL	8	0.6%
a2 cd 19 [-G]	2	0.2%
a2 cd 142 [T>C]	1	0.1%
a1 cd 59 [G>A]	1	0.1%
Total	1248	

Overall these results demonstrate extensive molecular heterogeneity underlying alpha thalassemia among the marginalized Kurds native to northern Iraqi Duhok City. The pattern includes globally prevalent alleles like -α3.7 and -α4.2 deletions, as well as region-specific point mutations like ααIVS1-5nt. However, frequencies vary considerably from central/southern Iraqi and even neighbouring Kurdish populations[29-31].

DISCUSSION

This study aimed to characterize the unique molecular epidemiology of alpha thalassemia among natives of Duhok City in the high-risk Iraqi Kurdish community. Using annotated genetic testing records spanning over nine years, our analysis determined the prevalent mutant alpha gene variants and their relative distributions in this population. Such in-depth understanding of population-specific mutations is vital to shape screening recommendations, prenatal diagnosis policies and counselling programs for thalassemia prevention[27, 32, 33].

The five most frequently encountered alpha globin genotype mutations in Duhok indigenous patients were homozygous or heterozygous -α3.7 and -α4.2 deletions, along with - -MED and ααIVS1-5nt alleles. This pattern agrees with earlier reports cataloguing common Middle Eastern variants, although proportions vary across Iraqi subpopulations[34]. Among our cohort, -α3.7 deletion displayed highest prevalence affecting over 43% alleles. The gene frequency aligns with observations by Al-Allawi et al[7]. and Hassanein et al.[34] in nearby Iraqi communities.

Considering alpha thalassemia demonstrates extensive molecular heterogeneity, the mutational spectra differs substantially based on geography and ethnicity[4]. Neighboring Iran exhibits high alpha globin gene mutation rates like Iraq, with -α3.7, - -MED and ααIVS-I-5nt similarly reported as common defects.[35-37] Conversely, Turkey harbors greater allelic diversity of less frequent variants besides these deletions[15,38,39]. Our results therefore enrich the scarce literature on alpha thalassemia genotyping among Iraqi Kurds. Duhok likely constitutes a genetically isolated community due to high endogamy even within this marginalized population[40]. The molecular patterns may thus diverge from Kurds residing in more cosmopolitan Iraqi cities or neighbouring nations.

The absolute numbers and annual frequency of mutant alleles displayed an upward trend from 2014-2022. Improved diagnostic modalities and wider testing penetration probably contributed rather than a genuine surge in incidence. Besides expected variation, we noted no drastic temporal fluctuations or emergence of new alleles over years. This tempo-spatial stability favors the postulate that Duhok Kurds represent an ancestrally homogeneous cluster. Collection of epidemiological indicators through national screening programs and registries will cement these initial surveillance insights.

Our analysis had limitations owing to the retrospective cross-sectional design restricted by archived record variables. We relied on testing undertaken for clinical indications rather than unbiased population sampling. Pediatric cases were overrepresented, while older adults often remain undiagnosed. Family history details were also inconsistently traced. Nevertheless, the study provides a unique long-term overview of alpha globin mutations in an understudied marginalized Iraqi community. Further local and regional collaborative efforts among clinicians, researchers and policymakers can now adopt these findings to inform

prevention policies targeting at-risk Kurdish subgroups.

Ongoing research can explore potential genotype-phenotype correlations associating specific alpha thalassemia mutations with clinical severity indicators. More consistent recording of demographic factors may also reveal population substructures linked to particular globin alleles. Systematic surveys should equally include larger adult representation through community-based recruitment. Ultimately a national-level thalassemia registry compiling mutational spectra and epidemiological trends can guide health policies aimed at disease control in Iraq's various high-risk ethnic enclaves like the Kurds.

CONCLUSION

This 9-year retrospective analysis of alpha thalassemia genotyping data offers seminal insights into the molecular epidemiology of mutant globin alleles among indigenous inhabitants of Duhok City in the Kurdish autonomous region of Iraq. The high carrier frequency and predominance of -α3.7 deletion mirrors broader Iraqi and Middle East Arab populations. However, the specific distribution of other common regional alleles like -α4.2, - -MED and ααIVS1-5nt varies significantly across ethnic and geographical subgroups. Our documentation serves as preliminary evidence guiding diagnostic workup, premarital/prenatal screening protocols and genetic counselling programs for thalassemia in this marginalized community. More vigorous surveillance and health promotion policies focused on Iraqi Kurds can help alleviate the substantial yet under-recognized disease burden in the region. Our documentation provides seminal evidence regarding site-specific mutational spectra to direct molecular screening protocols and guide targeted prevention policies for reducing thalassemia burden among the isolated Duhok community. Ongoing surveillance can monitor changing trends over upcoming years

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CONFLICT OF INTEREST

The author declared no conflicts of interest to disclose regarding this study.

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پوخته

ئەپیدێمیۆلۆجیای مۆلێکولی لە گۆڕانکارییەکانی ئالفا تالاسێمیا لە شاری دەۆک، هەرێمی کوردستانی عیراق: پێشبینینێکی 9 ساڵانە

پێشەکی و ئارمانج: تالاسیمیای ئەلفا بەشێوەیەکی بەرز لە عێراق بەلا دێتەوە، بەتایبەتی لەنێو گرووپە کوردییەکانی نزیکناسیی نێوخێوەیی. بەڵآم زانیاری کەم دەربارەی ئێپیدێمیۆلۆجیای مۆلێکولایی سەبەبداریی مووتاسیۆنی گلۆبین ئەلفا هەیە لەم نیفسەدا. ئەم لێکۆلینە هەڵوێست دەکات بۆ واڵمدانەوەی سپێکتروومی موتاسیۆن و بەشبوونی کاتی هێڵکەوتەکانی تالاسیمیای ئەلفا لەنێوان خەڵکی زێدەڕووی شاری دهۆکی عیراقی باکوور.

ڕێکێن کاری: لەم لێکۆلینە بینەرایی - بەشخستنەوەییدا، تۆماری تایبەت بە تالاسیمیای ئەلفای ژین بەرجەستەی ١٢٩٥ کەس لە دهۆک لە لابۆراتوارێکی تایبەتی عێراقی باکوور لە ٢٠١٤ تا ٢٠٢٢ زانیارییەکانی بۆ دیاریکردنی کردنی فریکوونسی جینۆتایپی مووتانت و ئەلیل شیکردنەوە.

ئەنجام: پێنج جینۆتایپی متحولی سەرەکی دۆزرایەوە بوون، (5.5%) -α3.7/-α3.7، -α3.7/αα (19.1%)، -αMED/αα (3.2%)، -α3.7/- αMED (2.6%) و ααIVS1-5nt/αα (1.5%). سڕینەوەی a3.7- باڵدەست بوو کە سەرجەم 43.8٪ ئەلیلی دەگرت. بەراورد بەهێزێکی ٩ ساڵانەی زیادبونەوەی ساڵانە دیار کرا.

دەستکەفتێن ڤەکولینێ: ئەم تایبەتمەندی مۆلێکوولاری حەشیمەتێکی سەرەتایی پێشکەش دەکات بۆ پێکهێنانی سیاسەتی سکرینینگ و پاراستن لە تالاسیمیا هەوێنەر بۆ کۆمەڵگای کوردی لاوازخراوی بەختەوەریی بەرز لە باکووری عێراق.

الخلاصة

تقييم فحص دوبلر (التخطيط فوق الصوتى) للدورة الجنينية المشيمية والدورة الرحمية المشيمية في النساء المصابات بتسمم ما قبل الولادة: المقارنة والارتباط بين مقايس الدوبلر المختلفة

الخلفية والأهداف: تواتر طفرات ألفا ثلاسيميا في مدينة دهوك، إقليم كردستان العراق: دراسة استرجاعية مدتها 9 سنوات الخلفية والأهداف: يظهر مرض ألفا ثلاسيميا معدل انتشار مرتفع في العراق، وخاصة بين المجموعات الفرعية الكردية ذات الأقارب. ومع ذلك، لا تزال البيانات محدودة فيما يتعلق بالوبائيات الجزيئية لطفرات ألفا جلوبين المسببة في هذه الفئة من السكان. هدفت هذه الدراسة إلى توضيح الطيف الطفري والتوزيع الزمني لأليلات ألفا ثلاسيميا بين السكان الأصليين لمدينة دهوك في شمال العراق.

الطرق: في هذه الدراسة المقطعية بأثر رجعي، تم تحليل سجلات الاختبار الجيني ألفا جلوبين الإيجابية لـ1295 من سكان دهوك الأصليين في مختبر متخصص في شمال العراق من 2014 إلى 2022 لتحديد النمط الجيني الطافر وترددات الأليل.

النتائج: الأنماط الجينية الطافرة الخمسة الأكثر شيوعا التي تم اكتشافها هي **(5.5%) -α3.7/-α3.7 ، -α3.7/αα (19.1%)، -αMED/αα (3.2%)، -αMED (2.6%) -α3.7/و-αIVS1-5nt/αα (1.5%).** كان الحذف a3.7- هو السائد مما يؤثر على 43.8% من الأليلات. ولوحظ وجود مسار كشف سنوي تصاعدي ثابت لمدة 9 سنوات.

الاستنتاجات: قد يوفر التحليل الجزيئي الخاص بالسكان أدلة أساسية لتشكيل سياسات فحص مرض الثلاسيميا والوقاية منه التي تستهدف المجتمع الكردي المهمش عالي الخطورة في شمال العراق.