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Parental Consanguinity and Chromosomal Abnormality Yield in a Five-Year Cytogenetic Referral Cohort from Eastern Libya

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زواج الأقارب ونسبة اكتشاف اختلال الصيغة الصبغية في مجموعة من المرضى المُحالين للتحليل الهيئة الكروموسومية خلال خمس سنوات في شرق ليبيا

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Abstract:

Background/Objective: Parental consanguinity is common in eastern Libya and is an established risk factor for autosomal-recessive disease, but its relationship to chromosomal abnormality is less clear; we tested whether consanguinity was associated with karyotype abnormality, reproductive loss, or previously affected children among patients referred to the First International Laboratory in Benghazi (2021–2025) from a 319-record cytogenetic registry. **Methods**: Consanguinity was coded as present, absent, or unknown; records with unknown status were excluded. The abnormality rate was compared between consanguineous and non-consanguineous referrals with an interpretable karyotype (Fisher's exact test), and reproductive history and parental age with the Mann–Whitney U and Fisher's exact tests. **Results**: Consanguinity was documented for 150 of 319 records (47.0%); 42 (28.0%) were consanguineous, mostly first-cousin unions. Among 107 patients with known consanguinity and an interpretable karyotype, a chromosomal abnormality was found in 11 of 26 (42.3%) consanguineous versus 37 of 81 (45.7%) non-consanguineous referrals (odds ratio [OR] 0.87, 95% CI 0.36–2.13; P = .82). Reproductive history did not differ: at least one prior miscarriage in 43.6% versus 39.8% (OR 1.17, 95% CI 0.55–2.46; P = .71), and at least one previously affected child in 26.7% versus 26.9% (OR 0.99, 95% CI 0.38–2.56; P = 1.00). Maternal and paternal age were also similar. **Conclusion**: Consanguinity was not associated with chromosomal abnormality yield or adverse reproductive history, consistent with consanguinity raising risk chiefly for autosomal-recessive single-gene disorders rather than aneuploidy; these findings support molecular rather than cytogenetic testing for consanguineous families with recurrent pregnancy loss.

Keywords: consanguinity, chromosomal abnormality, karyotype, reproductive loss, genetic counseling, Libya.

الملخص

الخلفية والهدف: زواج الأقارب شائع في شرق ليبيا ويُعدّ عامل خطر معروفًا للأمراض الوراثية المتنحية، غير أنّ علاقته باختلال الصيغة الصبغية (مقابل الاضطرابات أحادية الجين) أقلّ وضوحًا. هدفت هذه الدراسة إلى معرفة ما إذا كان زواج الأقارب مرتبطًا باختلال الصيغة الصبغية عند إجراء تنميط الصبغيات (الكاريوتايب)، أو بالخسارة الإنجابية ووجود أطفال مصابين سابقًا، بين المرضى المُحالين إلى المختبر الدولي الأول في بنغازي (2021–2025) ضمن سجلّ وراثي خلوي يضم 319 سجلًّا. طرق البحث: صُنّفت حالة زواج الأقارب إلى موجودة أو غائبة أو غير معروفة، واستُبعدت السجلات غير المعروفة الحالة. قُورنت نسبة الاختلال بين المُحالين

من ذوي زواج الأقارب وغيرهم ممن لديهم كاريوتايب قابل للتفسير (اختبار فيشر الدقيق)، وقُورن التاريخ الإنجابي وعمر الوالدين باختباري مان–ويتني وفيشر الدقيق. النتائج: وُثّقت حالة زواج الأقارب في 150 من 319 سجلًّا (47.0%)، وكان 42 منهم (28.0%) من ذوي زواج الأقارب، معظمهم من زواج أبناء العمومة من الدرجة الأولى. ومن بين 107 مرضى معروفي الحالة ولديهم كاريوتايب قابل للتفسير، وُجد اختلال الصيغة الصبغية لدى 11 من 26 (42.3%) من ذوي زواج الأقارب مقابل 37 من 81 (45.7%) من غيرهم (نسبة الأرجحية 0.87 [OR]، فاصل الثقة 95% 0.36–2.13 [CI]؛ قيمة الاحتمال 0.82 = [P]). ولم يختلف التاريخ الإنجابي باختلاف زواج الأقارب: إذ أُبلغ عن إجهاض سابق واحد على الأقل لدى 43.6% مقابل 39.8% (نسبة الأرجحية 1.17 [OR]، فاصل الثقة 95% 0.55–2.46 [CI]؛ قيمة الاحتمال 0.71 = [P])، وعن طفل مصاب واحد على الأقل لدى 26.7% مقابل 26.9% (نسبة الأرجحية 0.99 [OR]، فاصل الثقة 95% 0.38–2.56 [CI]؛ قيمة الاحتمال 1.00 = [P]). كما تشابه عمر الأم والأب. الاستنتاج: لم يرتبط زواج الأقارب بنسبة اختلال الصيغة الصبغية ولا بالتاريخ الإنجابي غير المواتي، بما يتّسق مع كون زواج الأقارب يرفع بشكل رئيس خطر الاضطرابات الوراثية المتنحية أحادية الجين لا اختلال الصبغيات العددي. وتدعم هذه النتائج اللجوء إلى الاختبارات الجزيئية بدلًا من الوراثية الخلوية لتقييم أسر ذوي زواج الأقارب التي تعاني من الإجهاض المتكرر.

الكلمات المفتاحية: زواج الأقارب، اختلال الصيغة الصبغية، تنميط الصبغيات، الخسارة الإنجابية، الاستشارة الوراثية، ليبيا.

Introduction

Parental consanguinity (marriage between blood relatives, most often first cousins) is a common social practice across the Middle East and North Africa, where an estimated one-fifth to one-half of marriages are consanguineous [1,2]. In eastern Libya, a survey of 699 married couples in Benghazi found a consanguinity prevalence of 46%, with first-cousin unions accounting for 40% of all marriages [3], broadly consistent with reports from neighboring Tunisia, Morocco, and Saudi Arabia [4,5,6]. An earlier hospital-based study in eastern Libya reported an association between consanguinity and congenital anomalies [7].

The clinical rationale for consanguinity screening is well established for autosomal-recessive single-gene disorders: related parents are more likely to carry the same rare recessive allele inherited from a common ancestor, and consanguinity substantially raises the recurrence risk for conditions such as hemoglobinopathies and inborn errors of metabolism [1,8]. Whether the same reasoning extends to chromosomal aneuploidy is less certain. Most autosomal and sex-chromosome aneuploidies arise from meiotic non-disjunction, a stochastic, largely maternal-age-driven process without an established recessive genetic basis, so a mechanistic link to consanguinity is not obvious [9]. Empirical findings have been mixed: some hospital-based series have reported a higher yield of chromosomal abnormality among consanguineous referrals [10], a large amniotic-fluid series from Iran found chromosomal abnormalities to be more frequent in non-consanguineous pregnancies [11], and a large infertility cytogenetic series from the United Arab Emirates found no significant association between consanguinity and chromosomal abnormality or polymorphism [12]. A preimplantation genetic testing study did find a higher rate of specific embryonic aneuploidies among consanguineous couples, but no difference in overall euploidy rate once maternal age was taken into account [13]. This heterogeneity suggests that any relationship between consanguinity and karyotype-detectable abnormality, if present, is modest and population-specific, in contrast to the well-replicated association between consanguinity and recessive disease.

Consanguinity status was recorded, albeit incompletely, throughout a 319-record cytogenetic registry at the First International Laboratory in Benghazi. Here we examine whether consanguinity was associated with the chromosomal abnormality rate on karyotyping, and whether consanguineous families reported more reproductive loss or previously affected children. We hypothesized, on the basis of the mechanistic distinction between recessive single-gene disease and meiotic non-disjunction, that consanguinity would show no association with chromosomal abnormality yield, reproductive loss, or previously affected children in this registry.

Materials and Methods

Study design, setting, and cohort

This retrospective study analyzed consanguinity data among patients referred for chromosomal analysis to the First International Laboratory, Faculty of Biomedical Sciences, University of Benghazi, between 2021 and 2025, drawn from a 319-record cytogenetic registry. The analysis is confined to consanguinity and reproductive-history variables. Records were anonymized before analysis and contained no patient-identifiable information. The overall cytogenetic spectrum of this registry and its Down syndrome subgroup, including cytogenetic subtype, age at diagnosis, and maternal age, are reported in companion analyses [16,17].

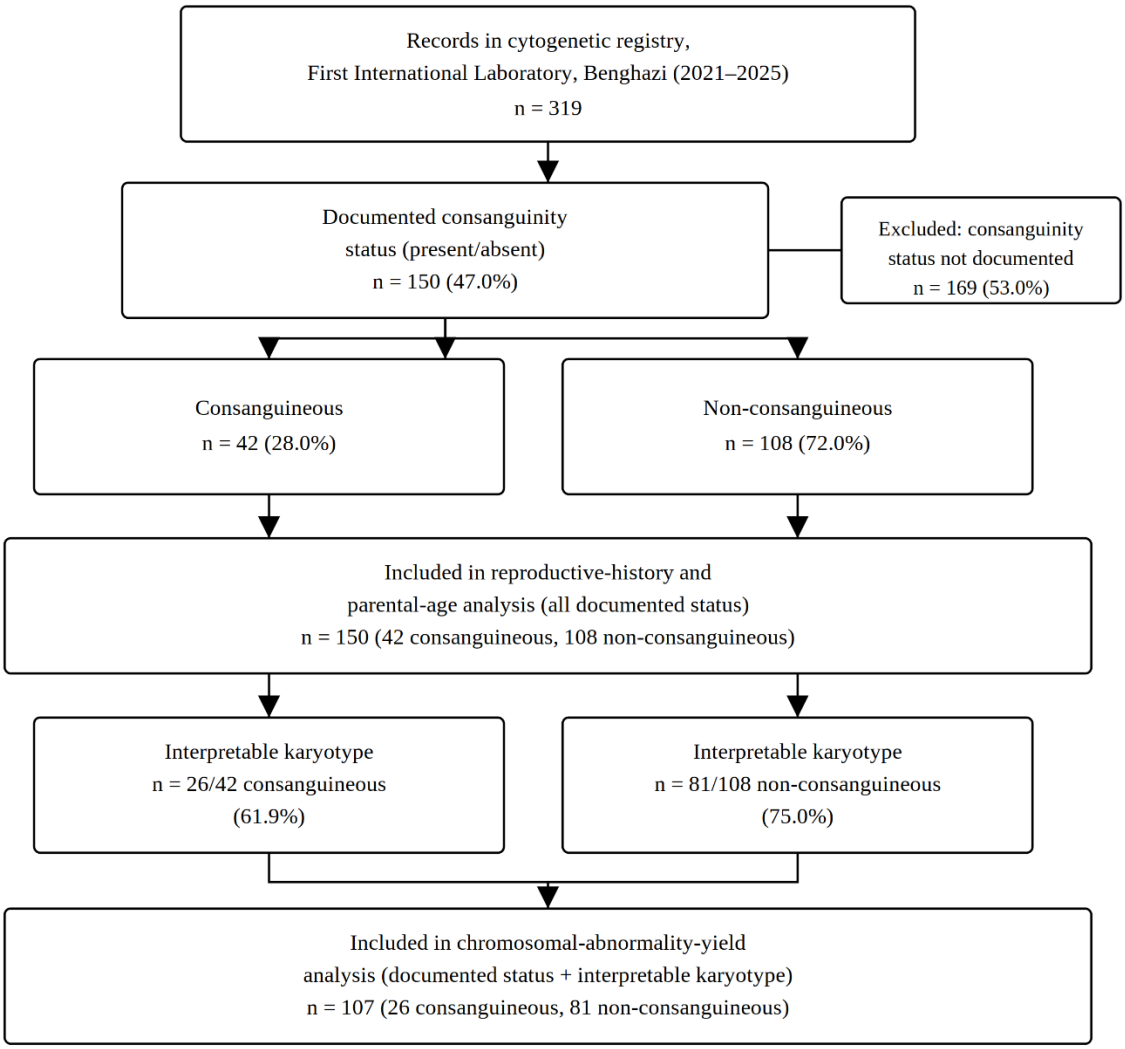

Figure 1. Study flow diagram: registry records, consanguinity-documentation status, and analytic subsets.

Consanguinity and reproductive-history variables

Parental consanguinity was coded conservatively as present, absent, or unknown from the referral record; ambiguous or numeric-only entries were coded as unknown rather than inferred. Where stated, the specific relationship (for example, first cousins) was retained as free text. Four reproductive-history variables were extracted where recorded: number of prior spontaneous abortions/miscarriages, number of siblings or prior children, number of previously affected (abnormal) children, and maternal and paternal age. These variables reflect family history and were analyzed regardless of the proband's own karyotype result.

Cytogenetic analysis and classification

Peripheral-blood lymphocytes were cultured for 72 hours in RPMI-1640 medium supplemented with fetal bovine serum, L-glutamine, penicillin–streptomycin, and phytohemagglutinin-M at 37 °C. Cell division was arrested with colcemid, followed by hypotonic 0.075 M potassium chloride treatment and fixation in 3:1 methanol–glacial acetic acid. Metaphase spreads were G-banded by trypsin–Giemsa (GTG) and described according to the International System for Human Cytogenomic Nomenclature (ISCN) [14]. Each karyotype was classified as normal, normal variant (benign heteromorphism), autosomal aneuploidy, sex-chromosome aneuploidy, structural abnormality, or other/complex, with mosaic cases flagged separately. A clinically significant chromosomal abnormality was defined as any category other than normal or normal variant.

Approximately 20 metaphase cells were examined per case at standard (non–high-resolution) banding resolution, and 3 to 5 metaphases were fully karyotyped. When mosaicism was suspected or detected, the number of cells examined was increased to 50.

Statistical analysis

The analysis used two denominators. For the chromosomal abnormality rate, the cohort was restricted to patients with both a documented consanguinity status (yes/no) and an interpretable karyotype (n = 107), since an abnormality rate cannot be computed for patients without a usable result. For reproductive-history and parental-age variables, all patients with documented consanguinity status were included regardless of karyotype outcome (n = 150), since these variables describe the family rather than the proband's own cytogenetic result. Categorical variables are summarized as counts and percentages with 95% Wilson confidence intervals (CIs) [18] and compared using Fisher's exact test [19], given the small cell counts in several strata. Continuous and count variables are summarized as median with interquartile range (IQR) and compared using the Mann–Whitney U test. A two-sided P value below .05 was considered statistically significant, and P values close to this threshold are described as borderline rather than significant. Analyses were performed using IBM SPSS Statistics version 26.0. For Mann–Whitney comparisons, an approximate rank-biserial correlation ($r = z/\sqrt{N}$) is reported alongside each P value as an effect-size estimate, back-calculated from the reported P value and sample size; by conventional benchmarks (0.10 small, 0.30 medium, 0.50 large), all fell in the small-to-negligible range.

Ethical considerations

The study used anonymized laboratory karyotype records that contained no patient-identifiable data. No individually identifiable information was accessed, and no clinical intervention was involved. Institutional permission to use the anonymized dataset for research purposes was granted by the laboratory (First International Laboratory, Benghazi; approval reference no. 26/91). Because the study used fully anonymized retrospective records containing no patient-identifiable information, informed consent was waived.

Results

Documentation and prevalence of consanguinity

Of 319 referred individuals, consanguinity status was documented as present or absent for 150 (47.0%); the remaining 169 (53.0%) had no usable entry and were coded as unknown and excluded from comparative analyses. Among the 150 with documented status, 42 (28.0%) were consanguineous and 108 (72.0%) were not. Where the degree of relationship was further specified (19 of the 42 consanguineous records, 45.2%), first-cousin unions predominated, consistent with the pattern reported for Benghazi in a household-based survey (46% consanguinity, 40% first cousins) [3]. Table 1 summarizes the characteristics of the two groups.

Table 1. Characteristics of patients with documented consanguinity status (n = 150).

Characteristic		Consanguineous (n = 42)	Non-consanguineous (n = 108)
Sex	Male	22 (52.4%)	52 (48.1%)
	Female	20 (47.6%)	54 (50.0%)
	Unknown	0 (0.0%)	2 (1.9%)
Age group	Neonate (<1 mo)	7 (16.7%)	24 (22.2%)
	Infant (1 mo–<2 y)	6 (14.3%)	24 (22.2%)
	Child (2–11 y)	8 (19.0%)	24 (22.2%)
	Adolescent (12–17 y)	4 (9.5%)	7 (6.5%)
	Adult (≥18 y)	17 (40.5%)	28 (25.9%)
	Unknown	0 (0.0%)	1 (0.9%)
Region	Cyrenaica (East)	35 (83.3%)	96 (88.9%)
	Fezzan/South	4 (9.5%)	6 (5.6%)
	Unknown	3 (7.1%)	6 (5.6%)
Interpretable karyotype obtained		26 (61.9%)	81 (75.0%)

Percentages are of the column total (consanguineous, n = 42; non-consanguineous, n = 108). Region and relationship detail were incompletely recorded, as noted in the Limitations.

Chromosomal abnormality yield by consanguinity status

Among the 107 patients with both documented consanguinity status and an interpretable karyotype, a clinically significant chromosomal abnormality was present in 11 of 26 consanguineous referrals (42.3%; 95% CI 25.5–61.1) and 37 of 81 non-consanguineous referrals (45.7%; 95% CI 35.3–56.5) (Fig. 2). This difference was not statistically significant (Fisher's exact P = .82; odds ratio [OR] 0.87, 95% CI 0.36–2.13, favoring neither group). The distribution of specific abnormality categories, shown in Table 2, was likewise similar between groups, although the small numbers in most categories preclude formal comparison beyond the overall abnormal/normal contrast.

Table 2. Cytogenetic classification by consanguinity status among karyotyped patients (n = 107).

Karyotype category	Consanguineous (n = 26)	Non-consanguineous (n = 81)
Normal	11 (42.3%)	44 (54.3%)
Normal variant (heteromorphism)	4 (15.4%)	0 (0.0%)
Autosomal aneuploidy (incl. mosaic)	6 (23.1%)	25 (30.9%)
Sex-chromosome aneuploidy (incl. mosaic)	2 (7.7%)	10 (12.3%)
Structural abnormality	2 (7.7%)	2 (2.5%)
Other/complex	1 (3.8%)	0 (0.0%)
Any clinically significant abnormality	**11 (42.3%)**	**37 (45.7%)**

Fisher's exact test for any abnormality vs. normal/normal variant: P = .82. 95% CI for the abnormal proportion: 25.5–61.1% (consanguineous) and 35.3–56.5% (non-consanguineous).

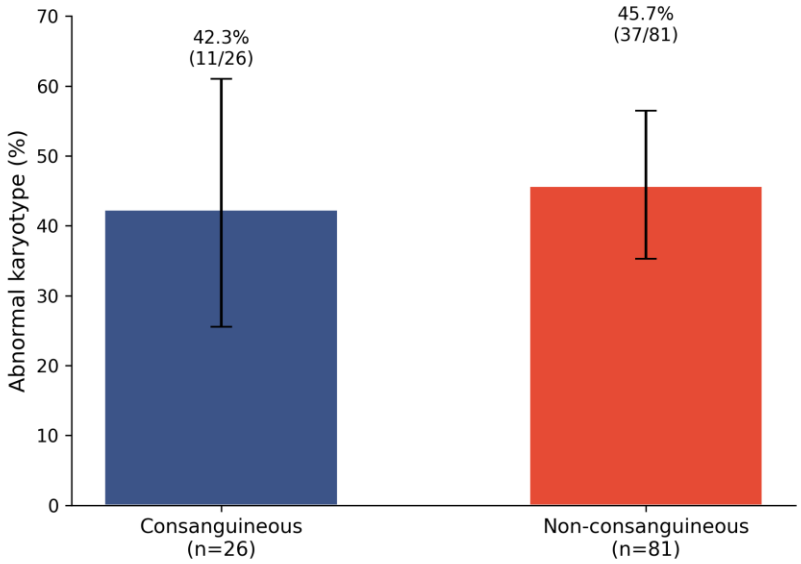

Figure 2. Chromosomal abnormality yield by consanguinity status, with 95% confidence intervals (n = 107).

Reproductive history and parental age

Among the 150 patients with documented consanguinity status, reproductive-history variables were available for 60–69% of records (Table 3). The median number of prior miscarriages was 0 in both groups (interquartile range [IQR] 0–2 among consanguineous families vs 0–1 among non-consanguineous families; Mann–Whitney P = .22, r = 0.10). At least one prior miscarriage was reported in 17 of 39 consanguineous families with data (43.6%) versus 41 of 103 non-consanguineous families (39.8%) (OR 1.17, 95% CI 0.55–2.46; Fisher's exact P = .71). The median number of siblings was slightly higher among consanguineous families (4 vs 3; P = .39, r = 0.07). The number of previously affected (abnormal) children did not differ by consanguinity (median 0 in both groups; P = .94, r = 0.01), and at least one previously affected child was reported at nearly identical proportions (8/30, 26.7% consanguineous vs 21/78, 26.9% non-consanguineous; OR 0.99, 95% CI 0.38–2.56; Fisher's exact P = 1.00) (Fig. 3). Maternal age (median 30 vs 33 years; P = .21, r = 0.11) and paternal age (median 36 vs 39 years; P = .13, r = 0.13) also did not differ significantly by consanguinity, although both were numerically lower among consanguineous couples. Age at referral of the proband was somewhat higher among consanguineous cases (median 11.0 years, IQR 0.8–31.5) than non-consanguineous cases (median 2.7 years, IQR 0.2–23.0; P = .053, r = 0.16), a difference that approached but did not reach conventional statistical significance.

Table 3. Reproductive history and parental age by consanguinity status (n = 150).

Variable	Consanguineous, median (IQR)	Non-consanguineous, median (IQR)	P value
Prior miscarriages (n avail.)	0 (0–2) [n = 39]	0 (0–1) [n = 103]	.22
Siblings/prior children (n avail.)	4 (1–5.5) [n = 37]	3 (1–5) [n = 101]	.39
Previously affected children (n avail.)	0 (0–1) [n = 30]	0 (0–1) [n = 78]	.94
Maternal age, years (n avail.)	30 (27–34.5) [n = 37]	33 (27–38) [n = 102]	.21
Paternal age, years (n avail.)	36 (31–42) [n = 37]	39 (33–46) [n = 97]	.13
Proband age at referral, years (n avail.)	11.0 (0.8–31.5) [n = 42]	2.7 (0.2–23.0) [n = 107]	.053

IQR, interquartile range (25th–75th percentile). P values from the Mann–Whitney U test. "n avail." denotes the number of records with a non-missing value for that variable. Approximate rank-biserial effect sizes (r) for these comparisons are given in the Results text and range from 0.01 to 0.16 (small to negligible).

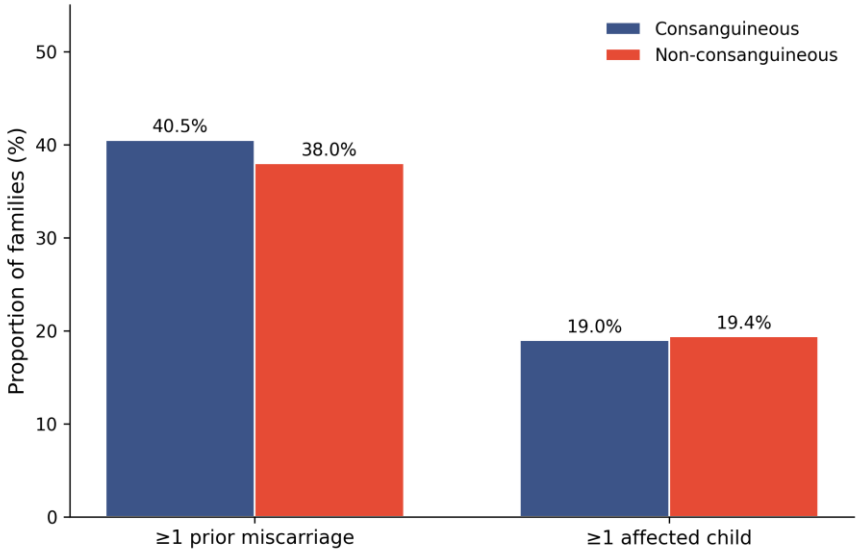

Figure 3. Proportion of families reporting at least one prior miscarriage or previously affected child, by consanguinity status.

Discussion

In this five-year cytogenetic registry from eastern Libya, parental consanguinity, documented in nearly half of referrals and present in more than a quarter of those with known status, was not associated with a higher yield of chromosomal abnormality on karyotyping, nor with more reproductive loss, more previously affected children, or younger parental age. Point estimates for abnormality yield were, if anything, numerically slightly lower among consanguineous referrals (42.3% vs 45.7%), and all reproductive-history comparisons crossed the null.

This null result is consistent with the mechanistic distinction noted in the Introduction: consanguinity elevates disease risk chiefly through shared recessive alleles [1,8], a route already linked to stillbirth and infant death in this population [15], whereas aneuploidy arises predominantly from maternal-age-related meiotic non-disjunction with no established recessive basis [9,20]. A large Egyptian clinical-genetics series likewise found consanguinity most strongly associated with autosomal-recessive disorders and least with chromosomal disorders [8]. Our finding agrees with the largest relevant comparison identified in the literature, a United Arab Emirates infertility cytogenetic series that found no significant consanguinity–chromosomal abnormality association [12], and with a large Iranian amniocentesis series in which abnormalities were, if anything, more frequent among non-consanguineous pregnancies [11]. It contrasts with smaller, older hospital-based series reporting higher abnormal-karyotype yields among consanguineous patients ascertained through Down syndrome and recurrent-miscarriage clinics [10], and with a preimplantation genetic testing study that found higher rates of specific chromosome-13/14 and chromosome-16 embryonic aneuploidies among consanguineous couples without an overall euploidy

difference once maternal age was accounted for [13]. Overall, any true effect of consanguinity on aneuploidy risk appears small, population-specific, and readily obscured by the stronger effect of maternal age, the pattern observed here.

The absence of an association with previously affected children is a more clinically important negative finding than it may first appear. Families are frequently referred for karyotyping, in this and other settings, specifically because consanguinity raises concern about recurrence in a subsequent pregnancy. Our data suggest that, for the karyotype-detectable abnormalities captured by this registry, consanguinity was not in fact predictive of having already had an affected child (26.7% vs 26.9%). This does not mean consanguineous families are free of elevated genetic risk; rather, conventional karyotyping is the wrong test to demonstrate or quantify that risk, which for consanguineous couples is concentrated in single-gene recessive disorders that require molecular or exome-based testing to detect [1,8,21]. This distinction has direct implications for genetic counseling: a consanguineous couple with a normal karyotype in an affected child should not be reassured that recurrence risk is low, since the relevant risk lies outside what a karyotype can show, a gap that structured premarital and preconception counseling programs elsewhere in the region have sought to close for other recessive conditions [22].

The borderline older age at referral among consanguineous probands (median 11.0 vs 2.7 years) was not a pre-specified hypothesis and should be interpreted cautiously given P = .053. If real, it could reflect that consanguineous families are more often referred later, for example through pediatric or adolescent evaluation of a suspected inherited syndrome rather than neonatal dysmorphology screening, but the registry's referral-indication field was too incompletely recorded to test this directly.

Limitations

Several limitations apply. Consanguinity was undocumented for 169 of 319 records (53.0%); if recording was non-random, the observed proportions and associations could be biased in either direction. The consanguineous, karyotyped subgroup was small (n = 26), so the study can exclude a large but not a small effect of consanguinity on aneuploidy yield. Consanguinity was usually recorded only as present or absent, without the degree of relationship that modifies recessive-disease risk. Reproductive-history variables were family-reported, missing for 31–40% of the cohort, and unverified. Finally, karyotyping cannot detect the submicroscopic and single-gene disorders most strongly linked to consanguinity; chromosomal microarray or exome sequencing would be required to evaluate that risk [23].

Conclusion

Parental consanguinity, present in more than a quarter of referrals with documented status in this eastern Libyan cytogenetic registry, was not associated with a higher rate of chromosomal abnormality on karyotyping, nor with increased reproductive loss or previously affected children. These findings align with the mechanistic expectation that consanguinity chiefly elevates risk for autosomal-recessive single-gene disease rather than de novo chromosomal non-disjunction, and they argue against using karyotype results to reassure or alarm consanguineous families about recurrence risk. Routine, structured recording of consanguinity status and relationship degree would strengthen future registry-based studies, and consanguineous families with recurrent pregnancy loss or an affected child are better served by molecular or exome-based recessive-disease testing than by karyotyping alone.

Declarations

Ethics approval and consent. Institutional permission to use the anonymized dataset was granted by the laboratory (First International Laboratory, Benghazi; approval reference no. 26/91). Informed consent was waived owing to the use of fully anonymized retrospective records containing no patient-identifiable information.

***Availability of data*.** The anonymized dataset supporting this study is available from the corresponding author on reasonable request.

Competing interests. The authors declare no competing interests.

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***Author contributions*.** A. Abusneina: conceptualization, data curation, formal analysis, visualization, and writing, original draft. T. M. Shoeib: resources, review and editing. Both authors read and approved the final manuscript.

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