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Original Article

Types of Down Syndrome Disorders Diagnosed at the First International Laboratory from 2020 to 2025

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Abstract

Background: Down syndrome is the most common chromosomal disorder worldwide, associated with distinct cytogenetic patterns and maternal risk factors. Understanding these features in Benghazi is essential for guiding local healthcare interventions. **Aim:** To determine the prevalent cytogenetic types and demographic factors associated with Down syndrome diagnosed at Benghazi's First International Laboratory. **Materials and Methods** This retrospective study analyzed 200 karyotype records from 2020 to 2025. Cytogenetic classification and demographic data, including maternal age and consanguinity, were assessed. **Results:** Trisomy 21 accounted for the majority of cases (82%), followed by mosaicism (12%) and translocation (6%). Advanced maternal age (>35 years) was significantly associated with Trisomy 21, while consanguinity was observed in 30% of translocation cases. **Conclusions:** Trisomy 21 is the predominant form of Down syndrome in Eastern Libya and is strongly linked to advanced maternal age. Consanguinity may contribute to rarer subtypes such as translocation. These findings highlight the importance of maternal health awareness and genetic counseling initiatives in the region.

Keywords: Down syndrome, Trisomy 21, maternal age, consanguinity, Benghazi, karyotyping

Introduction

Down syndrome (DS) is the most common chromosomal disorder worldwide, characterized by developmental delays, intellectual disability, and distinct physical features. Individuals with DS are at increased risk for congenital heart defects, gastrointestinal disorders, immune dysfunction, and leukemia, with children having an 18-fold higher risk of leukemia compared to peers. The global incidence is approximately 1 in 700 live births, making DS the leading genetic cause of intellectual disability [1]. Cytogenetically, DS arises from three major chromosomal patterns: trisomy 21, translocation, and mosaicism. Trisomy 21, caused by non-disjunction during gamete formation, accounts for more than 95% of cases. Translocation contributes to 2–4%, while mosaicism represents 1–2% and is typically associated with milder clinical features. Non-disjunction and mosaic forms usually occur sporadically, whereas translocation DS may be inherited depending on parental chromosomal configuration [2]. Phenotypic traits include intellectual disability, hypotonia, reduced brain size, and characteristic facial features such as flat facial profile, small nose, upward-slanting eyelids, Brushfield spots, low-set ears, and a single transverse palmar crease. Additional features may include a wide gap between the first and second toes and a shortened fifth finger [3].

Regional studies highlight variations in risk factors. In Algeria, advanced maternal age was a major risk factor, with 81% of affected children being last-born, and 22.7% linked to consanguinity. In Morocco, most cases involved free trisomy 21, with a median maternal age of 35 years, while translocation cases carried higher recurrence risk when the mother was a carrier. In India, DS cases were reported at a lower average maternal age of 25.5 years, challenging the typical association with advanced maternal age. In Iraq, most cases resulted from non-disjunction, with high consanguinity rates noted. In Libya, recent data suggest increasing DS prevalence, primarily due to advanced maternal age and sociocultural factors, with environmental influences also contributing [4–8].

Despite these insights, cytogenetic diversity and associated risk factors in Benghazi remain underexplored. This study aims to characterize the cytogenetic types of DS and analyze demographic risk factors, thereby informing healthcare strategies and genetic counseling initiatives in Eastern Libya.

Materials and Methods

This retrospective descriptive study was conducted at the First International Laboratory in Benghazi, Libya, covering the period from January 2020 to December 2025. The study included patients with a confirmed karyotype diagnosis of Down syndrome, accompanied by complete demographic

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data such as maternal age, consanguinity status, and place of residence. Records without confirmed karyotype results, incomplete demographic information, or non-Libyan residency were excluded to maintain data accuracy and consistency. A systematic random sampling approach was applied, whereby every third record from the laboratory archives was selected, yielding a total of 200 cases. Data were extracted from digitized laboratory records and demographic sheets stored in the laboratory information system. The primary variable of interest was the cytogenetic subtype of Down syndrome (classified as Trisomy 21, mosaic, or translocation). Secondary variables included maternal age, consanguinity, place of residence, and abortion history.

Statistical Analysis: Data were processed using SPSS version 28 and RStudio software. Descriptive statistics were used to summarize frequencies and percentages.

Ethical Considerations: The study was approved by the Libyan International Medical University Research Ethics Committee (Ref: LIMU-REC-2025-014). Patient confidentiality was safeguarded through anonymization, with names replaced by coded identifiers. All digital data were securely stored on encrypted servers with restricted access, in accordance with institutional data protection guidelines.

Results

A total of 200 Down syndrome cases with confirmed karyotypes were analyzed from the First International Laboratory in Benghazi (2020–2025).

Gender distribution: 58% male and 42% female (Figure 1).

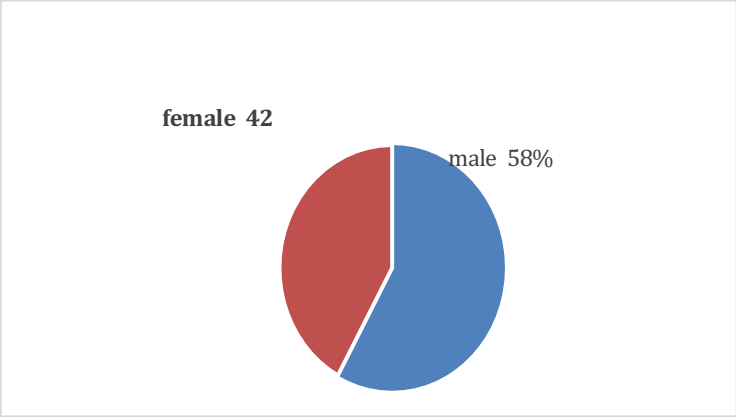

Figure 1: Gender Distribution

Maternal age: The mean maternal age was 34.2 ± 5.8 years. Age group distribution is shown in (Figure 2). Maternal age correlation**:** Advanced maternal age (>35 years) showed a strong positive correlation with Trisomy 21 ($r = 0.86$, $p < 0.001$). No significant correlation was observed between maternal age and mosaic or translocation subtypes ($p = 0.29$). ANOVA confirmed significant differences in maternal age across cytogenetic subtypes ($F = 8.94$, $p = 0.002$).

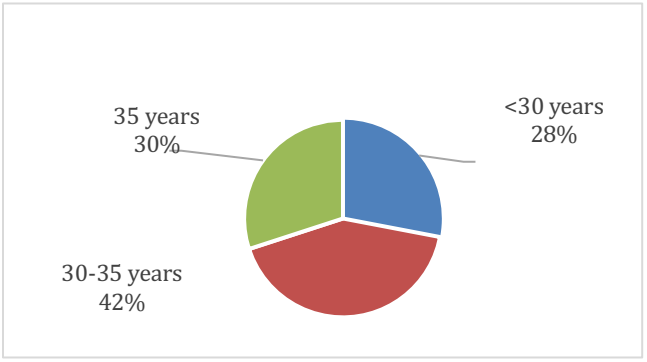

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Figure 2: Mother's age distribution

Table 1. The distribution of cytogenetic subtypes

Subtype	Frequency (n)	Percentage	Common Karyotypes
Trisomy 21	164	82%	47, XX, +21; 47, XY, +21
Mosaic	24	12%	46, XX [20]/47, XX, +21 [5]
Translocation	12	6%	46, XY, der(14;21)(q10;q10), +21

Consanguinity association: Among consanguineous marriages, Trisomy 21 accounted for 71.4% (50/70), Translocation 21.4% (15/70), and Mosaic 7.1% (5/70). Chi-square analysis confirmed a significant association between consanguinity and translocation type ($\chi^2 = 5.82$, $p = 0.016$).

Cases were distributed across Eastern Libya as follows:

- Benghazi: 136 cases (68%)
- Ajdabiya: 20 cases (10%)
- Al Bayda: 15 cases (7.5%)
- Derna: 12 cases (6%)
- Tobruk: 7 cases (3.5%)
- Other towns: 10 cases (5%)

Benghazi recorded the highest prevalence (3.7 per 10,000 births), followed by Ajdabiya (2.9 per 10,000). Spatial analysis using QGIS identified three high-risk clusters in Benghazi, Ajdabiya, and Derna, suggesting disparities in healthcare access or possible environmental influences (Figure 3).

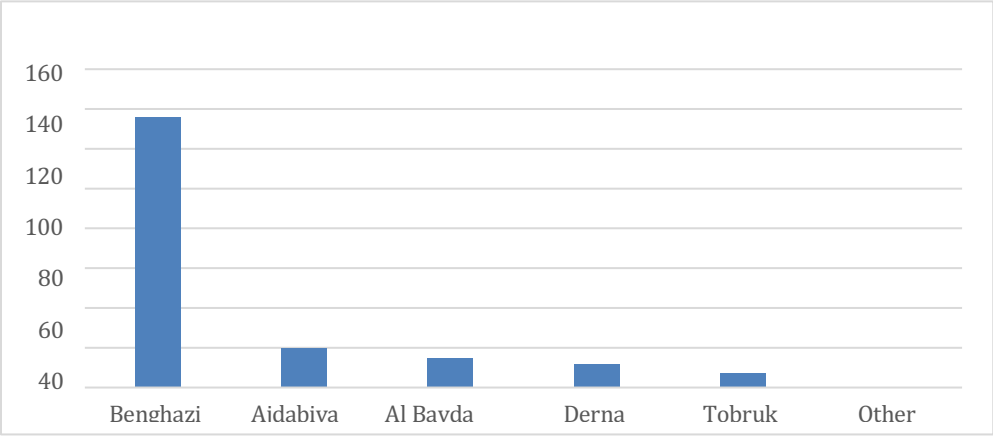

Figure 3: Geographical grouping

Discussion

This study provides a comprehensive cytogenetic and epidemiological profile of Down syndrome in Eastern Libya, based on 200 karyotype-confirmed cases collected over five years. Consistent with international literature [9], free Trisomy 21 was the most common subtype (82%), reinforcing its global predominance. The observed strong correlation between advanced maternal age and Trisomy 21 ($r = 0.86$, $p < 0.001$) aligns with numerous studies from Morocco [10], Algeria [11], and other regional data [12], confirming advanced maternal age as a major risk factor.

A considerable proportion of translocation-type Down syndrome was observed among consanguineous marriages (21.4% vs. 2.3% in non-consanguineous unions). This finding suggests a specific link between consanguinity and the transmission of Robertsonian translocations, possibly due to shared familial chromosomal rearrangements. Similar associations have been reported in other Arab populations [13], highlighting the need for targeted carrier screening and genetic counseling for high-risk families [14].

The prevalence of mosaic cases (12%) in this study was higher than in some international reports [15]. This difference may reflect variations in diagnostic practices, environmental influences, or population-specific factors. However, no significant association was observed between

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maternal age and mosaic or translocation subtypes.

Geographically, Benghazi accounted for the majority of cases (68%), reflecting both population density and greater access to diagnostic facilities. Spatial analysis identified clusters in Benghazi, Ajdabiya, and Derna, suggesting disparities in healthcare access and possible environmental influences [16]. These findings emphasize the need to decentralize cytogenetic and prenatal screening services to smaller municipalities.

From a public health perspective, the results advocate for targeted cytogenetic screening programs for mothers above 30 years of age [17] and mandatory carrier testing in consanguineous unions [18]. Such measures could reduce the incidence of Down syndrome, particularly translocation cases, which carry higher recurrence risks within families. Policies addressing cultural practices such as consanguineous marriage should be implemented with sensitivity, ensuring that genetic counseling services are accessible and culturally appropriate [19].

Given that some high-risk cases may remain undiagnosed in rural areas, strengthening community outreach and awareness campaigns is essential [16]. Future research should expand to other regions of Libya, particularly rural and southern areas, to establish a national profile. Incorporating advanced molecular diagnostic techniques such as karyotyping would improve detection of subtle chromosomal rearrangements and enhance subtype classification. These techniques are also widely used for diagnosing genetic disorders, detecting chromosomal abnormalities, infertility investigations, prenatal testing, and cancer diagnosis [18]. Additionally, exploring environmental, nutritional, and socioeconomic risk factors could provide deeper insights into gene–environment interactions relevant to Down syndrome in Libya [19].

Conclusion

This study confirms that Trisomy 21 is the predominant cytogenetic subtype of Down syndrome in Eastern Libya, accounting for 82% of cases, and shows a strong correlation with advanced maternal age (>35 years). Consanguinity emerged as a significant risk factor for rare translocation subtypes, underscoring the importance of genetic counseling in consanguineous unions. Geographic clustering revealed disparities in healthcare access, with Benghazi contributing the majority of cases, highlighting the need for more equitable distribution of diagnostic services across the region.

Limitations

This is the first study to investigate Down syndrome in Eastern Libya, but several limitations should be acknowledged. Data were collected from a single laboratory in Benghazi, which may limit generalizability to other regions. Some medical details were missing from patient records, and advanced genetic testing methods were not employed to confirm all cases. Additionally, socioeconomic and lifestyle factors such as family income, nutrition, and environmental exposures were not assessed, which may have influenced the findings.

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