RESEARCH PAPER

Chromosomal Study in Patients with Intellectual Disability: A Tertiary Care Hospital Study in North-East India.

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Abstract

Introduction: Chromosomal abnormalities, specifically numerical aneuploidies such as Trisomy 21, are significant contributors to intellectual disability globally, affecting approximately 2.5–3.0% of the population. This study aimed to evaluate the prevalence and nature of chromosomal abnormalities in patients with severe ID in a tertiary care setting in North-East India.

Methods: A hospital-based observational study was conducted on 30 clinically diagnosed patients with severe ID. Peripheral blood lymphocyte cultures were performed using RPMI-1640 medium, followed by G-banded karyotyping. At least 20 metaphase spreads were analyzed per patient to exclude mosaicism, following standard cytogenetic protocols.

Results: Chromosomal abnormalities were identified in 10% (3/30) of the cases. All abnormal cases were confirmed as free Trisomy 21 (47, XY,+21), which is the most frequent autosomal aberration in similar cohorts.
A correlation with advanced maternal age (mean >33 years) was observed in the positive cases.

Conclusions: Cytogenetic screening remains an essential tool for diagnosing the ethology of severe ID. Early identification of Trisomy 21 is critical for clinical management and providing accurate genetic counselling regarding recurrence risks in subsequent pregnancies.

Keywords: Intellectual disability, Cytogenetics, Karyotyping, Down syndrome, Trisomy 21, Chromosomal abnormalities.

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Introduction

Intellectual disability is a major global health concern, characterized by inadequate development of mental capacities and associated behavioural challenges [1]. In India, genetic and congenital malformations are a significant cause of infant and childhood mortality, with ahigh disease burden recorded in various population screenings [2]. Chromosomal abnormalities are identified as a primary ethology, contributing to approximately 15% of severe ID cases[1] . Down syndrome, resulting from Trisomy 21, is the most prevalent autosomal aneuploidy in live births [3]. While free trisomy accounts for the vast majority (92–95%) of cases, other variants such as Robertsonian translocation and mosaicism present different recurrence risks[4]. Although India reports approximately 21,000 Down syndrome births annually, there is a lack of localized data from the North-Eastern region[5]. This study was designed to evaluate patients in a tertiary care setting to provide accurate cytogenetic diagnoses and facilitate parental counseling.

Materials and Methods

This observational study was conducted at Multi-disciplinary Research Unit, J.M.C & H, Jorhat a Tertiary Care Hospital in North-East India. Ethical clearance and informed consent were obtained for all participants. It was done with the correlation of Department of Psychiatry,
J.M.C & H, Jorhat.

Laboratory Procedure:

1. **Sample Collection:** 3 mL of peripheral venous blood was collected in heparinized vacutainers.
2. **Culture:** Lymphocytes were cultured in RPMI-1640 medium supplemented with 10% fetal bovine serum and phytohemagglutinin, a powerful mitogen for T-cells, at 37°C for 72 hours [6].
3. **Harvesting:** Metaphase arrest was induced using colchicine to obtain a high yield of mitotic cells. This was followed by hypotonic treatment (0.075 M KCl) and fixation with a methanol-acetic acid (3:1) solution [7].
4. **Analysis:** Slides were prepared via the air-drying technique and subjected to GTG banding [6]. Karyotypes were analysed under light microscopy, examining 20 metaphase spreads per case to ensure the exclusion of mosaicism.

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Results

Of the 30 patients screened, 3 (10%) exhibited chromosomal abnormalities. All three cases were identified as free Trisomy 21 with a 47,XY,+21 karyotype.

- **Phenotypic Correlation:** All cytogenetically positive cases displayed classical features of Down syndrome, including facial dysmorphism, epicanthal folds, and developmental delay.
- **Maternal Age:** In the identified cases, the maternal ages at conception were 33, 35, and 35 years. This aligns with findings that maternal age-dependent factors significantly increase the risk of chromosomal nondisjunction [8].
- **Structural Variants:** No cases of Robertsonian translocation or mosaicism were identified in this specific cohort.

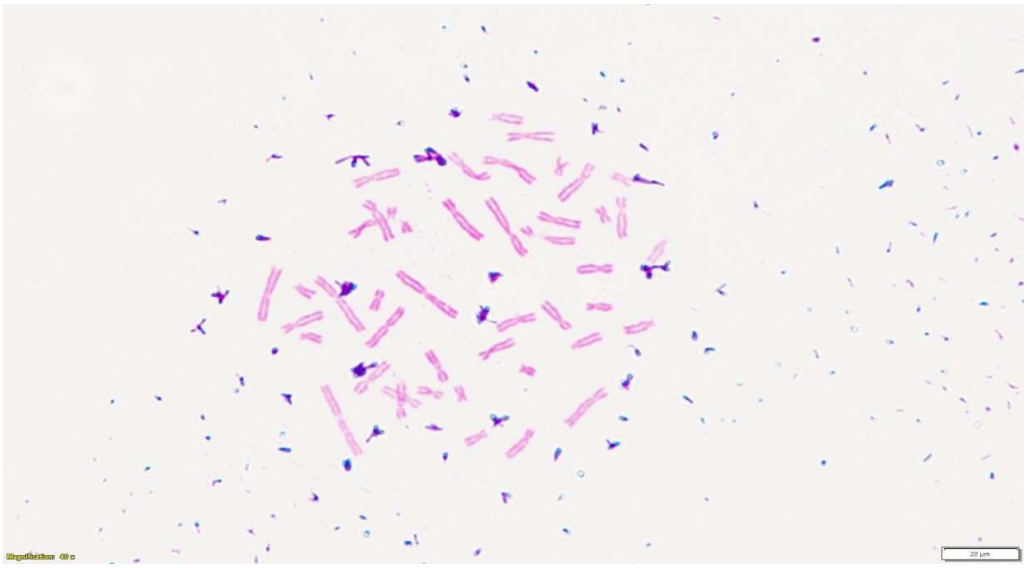

Figure -1: Metaphase Spread Free Trisomy 21

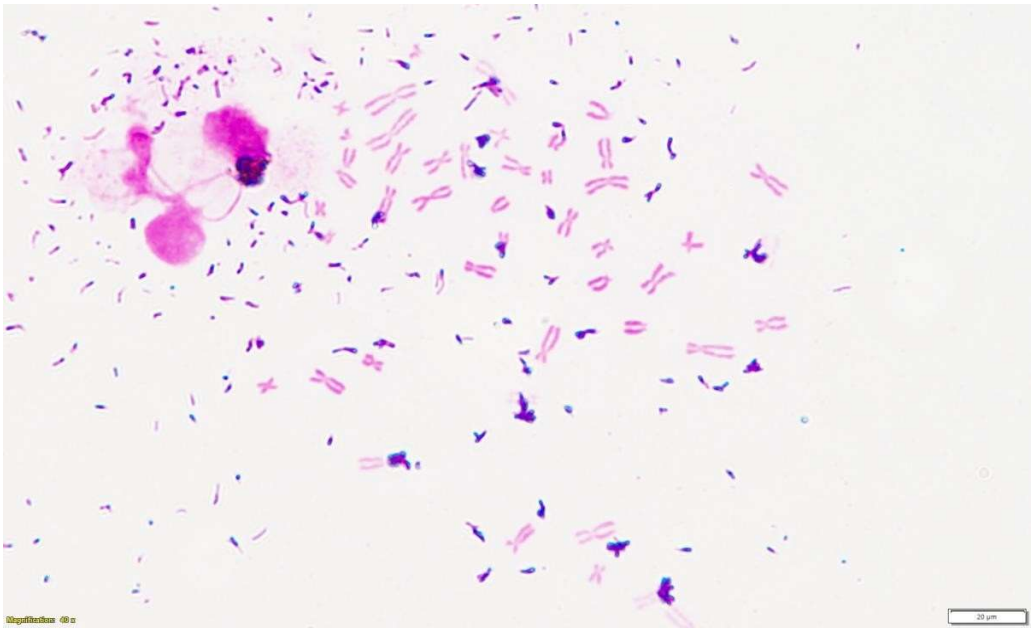

Figure -2: Metaphase Spread Free Trisomy 21

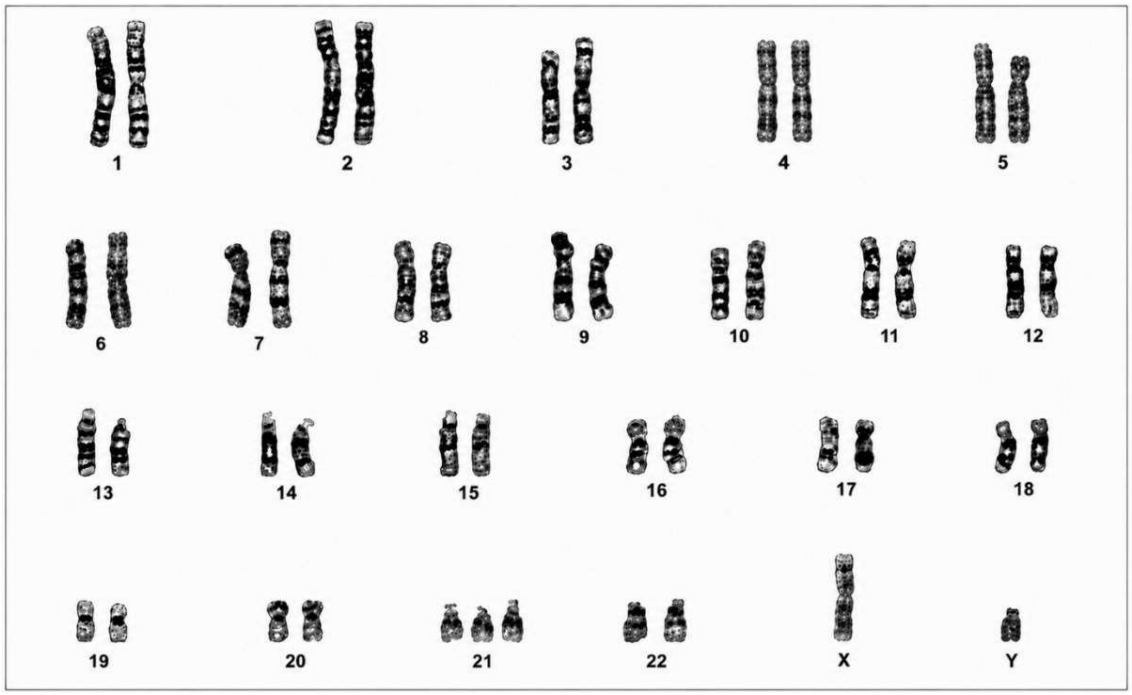

Figure -3: Ideogram Free Trisomy 21

Discussion

The findings of this study reinforce the role of numerical chromosomal abnormalities in the pathogenesis of severe ID. The 10% detection rate of Trisomy 21 in this cohort is consistent with Indian studies where Down syndrome is frequently reported as the most common chromosomal abnormality [9,10]. The identification of free Trisomy 21 as the sole abnormality in this group matches broader trends in India, where free trisomy often exceeds 90% of all Down syndrome cases [11, 4]. The observed correlation with maternal age (>33 years) supports the established link between advanced maternal age and meiotic nondisjunction errors [8]. Early cytogenetic diagnosis is not merely a tool for clinical confirmation but is essential for rehabilitation planning and providing accurate recurrence risk assessments for families [3]. In regions like North-East India, where specialized genetic services may be limited, routine karyotyping remains a robust and indispensable diagnostic approach.

Conclusion

G-banded karyotyping remains a gold-standard diagnostic tool in the evaluation of intellectual disability. This study identified free Trisomy 21 as the primary chromosomal abnormality in the screened cohort. Integrating routine cytogenetic screening into public health services is recommended to improve clinical management and facilitate genetic counselling for affected families in North-East India.

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