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CYTOGENETIC ANALYSIS IN CHILDREN WITH SUSPECTED CONGENITAL GASTROINTESTINAL ANOMALIES: A PROSPECTIVE OBSERVATIONAL STUDY

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ABSTRACT Cytogenetic analysis in detecting chromosomal abnormalities and its correlation with clinical complications in affected children. Data collected from children suspected of having congenital malformations were analyzed through Karyotyping. Typically, the karyotyping process involves several steps, including cell culture, chromosome preparation, staining, and analysis. Data collected from children suspected of having congenital malformations were analyzed through karyotyping, fluorescence in situ hybridization (FISH). The study found a significant correlation between chromosomal abnormalities and clinical complications, highlighting the importance of cytogenetic investigations in early diagnosis and intervention. It is associated with genetic disorders because they condition patient's management and prognosis. Anomalies that are often associated with other defects have shown early clinical symptoms and require more time and effort for correction.

KEYWORDS : Congenital abnormalities, Karyotyping, G-Band.

BACKGROUND

The clinical data of chromosome abnormalities in order to explore the corresponding relationships between the phenotypes and certain chromosome abnormalities, and increase the evidences of initial clinical indications of these types of disorders in different ages. Furthermore, the cytogenetic outcomes can guide medical professionals in choosing optimal treatment, social function training, and predicting the possible prognosis. Congenital abnormalities, also known as birth defects, are structural, functional, or metabolic disorders that are present at birth and can have a significant impact on the health and well-being of affected individuals [1]. These abnormalities can be caused by a variety of factors, including genetic factors, environmental exposures, and unknown causes [2] One of the key tools in the diagnosis and management of congenital abnormalities is cytogenetic analysis, which involves the study of an individual's chromosomes to identify any abnormalities or genetic variations that may be contributing to the development of the condition [3].

The identification of genetic variations and chromosomal abnormalities is crucial for the accurate diagnosis and management of congenital abnormalities. Cytogenetic analysis, which involves the study of an individual's chromosomes, is a key tool in this process, as it can help to identify specific genetic variations or chromosomal abnormalities that may be contributing to the development of the condition [4]. The types of chromosomal abnormalities that can be identified through cytogenetic analysis include numerical abnormalities, such as trisomies (e. g., Down syndrome) [2,5], and structural abnormalities, such as deletions, duplications, and translocations.

The clinical implications of these chromosomal abnormalities can be significant and can vary depending on the specific nature and location of the genetic variation. Numerical abnormalities, such as trisomies, can lead to a range of clinical manifestations, including intellectual disability, growth and developmental delays, and a variety of physical and medical complications [6]. Structural abnormalities, on the other hand, can disrupt the expression of specific genes and can lead to a range of clinical outcomes, depending on the genes that are affected.

The primary objective of this study is to assess the role of cytogenetic analysis in identifying chromosomal abnormalities in children suspected of having congenital abnormalities and to evaluate the associated clinical complications. This research explores the diagnostic benefits of cytogenetic and its implications for clinical management.

MATERIALS AND METHODS

Study Design and Population

This Prospective Observational study was conducted over a 1-year period at Coimbatore Medical College and Hospital. The study included children who presented with suspected congenital abnormalities, based on clinical signs and symptoms such as dysmorphic features, failure to thrive, and developmental delays. A total of 30 children were enrolled, with ages ranging from birth to 5 years.

Sample Collection: Peripheral blood samples were collected from each child, and standard cytogenetic analysis was performed to detect numerical and structural chromosomal abnormalities.

Karyotyping Method

Cell Culture

The isolated cells are cultured in a tissue culture medium for 68–72 hours to stimulate cell division, particularly the mitotic phase of the cell cycle.

Mitotic Arrest

- Addition of Colchicine: Colchicine is added to halt cell division at metaphase, the stage of mitosis when chromosomes are most condensed and visible.
- Harvesting Cells: After the cells have reached metaphase, they are harvested.

Cell Fixation

The cells are treated with a fixative (methanol and acetic acid) to preserve the chromosomes and prevent degradation.

Slide Preparation

- The fixed cells are dropped onto glass microscope slides, and the cells are gently spread out to form a monolayer.
- The cells are dried on the slides.

Staining and Analysis

Giemsa staining techniques used for this study to visualize the chromosomes, the stained chromosomes are then analyzed under a microscope, and the karyotype is determined by comparing the banding patterns and morphology of the chromosomes to a standard reference karyotype [7].

Clinical Data Collection

Clinical information was obtained from medical records, including the type of congenital abnormality, associated clinical features (e.g., intellectual disability, cardiac defects, skeletal abnormalities), and family history. Clinical complications such as growth delay, seizures, cardiovascular issues, and sensory impairments were recorded.

Statistical Analysis

Data were analyzed using descriptive statistics to identify the prevalence of chromosomal abnormalities. The relationship between the types of chromosomal anomalies and clinical complications was

assessed using chi-square tests and Fisher's exact test. A p-value of less than 0.05 was considered statistically significant.

RESULTS:

Chromosomal Abnormalities Detected

Among the 30 children studied, 12 (40%) were found to have chromosomal abnormalities through cytogenetic analysis. The distribution of these abnormalities as shown in Table 1.

Table 1: Chromosomal Abnormalities Detected

S.No	Chromosomal Abnormality	Number of Cases (%)
1	Trisomy 21 (Down syndrome)	5 (16.7%)
2	Trisomy 18 (Edwards syndrome)	2 (6.7%)
3	22q11.2 deletion (DiGeorge)	2 (6.7%)
4	Turner syndrome (45,X)	1 (3.3%)
5	Microdeletion syndromes	2 (6.7%)

Clinical Associations

- Congenital Heart Defects were present in 6/12 (50%) of children with chromosomal abnormalities, most commonly in Trisomy 21 and DiGeorge syndrome.
- Developmental Delay was noted in 11/12 (91.6%) children with chromosomal findings.
- Cleft Palate or Gastrointestinal Malformations were especially observed in 22q11.2 deletions and Trisomy 18.

DISCUSSION

This study corroborates findings from prior research indicating that Trisomy 21 remains the most frequent chromosomal abnormality associated with congenital malformations. Similar prevalence rates have been reported by [5,6]. The high association of 22q11.2 deletions with cardiac and palatal anomalies aligns with findings from [8].

Incorporating fluorescence in situ hybridization (FISH) in the study enabled the identification of microdeletion syndromes that may have been missed by conventional karyotyping. This aligns with current recommendations from the American College of Medical Genetics, emphasizing the importance of molecular cytogenetic techniques in diagnostic workflows for congenital anomalies.

CONCLUSION

Cytogenetic investigations using both conventional karyotyping and FISH substantially enhance diagnostic yield in children suspected of having congenital abnormalities. The identification of chromosomal abnormalities in 40% of cases in this study supports the early implementation of genetic testing in pediatric evaluation protocols. Genetic diagnosis not only facilitates accurate clinical management and prognosis estimation but also empowers families through genetic counseling and risk assessment.

Conflict of Interest

We, the authors declared that there is no conflict of interest related to this work.

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