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Case Report

Trisomy 18 Chronicles: A Case Report Illuminating Edward Syndrome

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ABSTRACT

This case report delves into the complexities surrounding Edwards syndrome (Trisomy 18), a chromosomal abnormality stemming from meiotic disjunction. Initially reported in 1960, Edwards syndrome is the second most prevalent autosomal trisomy following Down syndrome, affecting between 1 in 3500 and 1 in 7000 individuals, with a slight male predominance. The majority of cases involve parents under 30 years old. A review of 152 cases demonstrates a myriad of anomalies, including severe intellectual deficits, congenital heart problems and distinct facial features. This report aims to enhance understanding of Edwards syndrome by presenting a comprehensive case study detailing the diagnosis and distinctive clinical features of a newborn with Trisomy 18. This case report helps in understanding the complexity of genetics and clinical manifestations, highlighting the importance of prenatal diagnosis and counseling, multidisciplinary care approaches, parental decision-making, palliative care and the need for further research. The case report underscores the necessity for heightened public awareness and support for affected families. This study contributes to the body of knowledge regarding Trisomy 18, fostering a more compassionate and informed approach to managing this challenging genetic disorder, ultimately aiming to improve the quality of life for those affected by Edwards syndrome.

Keywords: Trisomy 18, Edwards Syndrome, Chromosome, Genetic Screening, Karyotype

INTRODUCTION

Meiotic disjunction, an error occurring during cell division, serves as the fundamental mechanism behind the development of Edward's syndrome[1]. Initially reported by Edwards et al. and Smith et al. [2,3] in 1960, This autosomal trisomy ranks as the second most common, trailing behind Down syndrome in prevalence. Edward syndrome can impact individuals at a frequency ranging from 1 in 3500 to 1 in 7000. The male-to-female ratio stands at 1.3:1. In over half of the instances, both parents are under the age of 30. A study encompassing 152 cases of Edward syndrome has been conducted, employing cytogenetic analysis to provide substantiation [4]. A wide range of anomalies, such as severe intellectual deficits, congenital heart problems, and unique facial traits, are among the clinical signs of trisomy 18[5]. The most common issues concerning the cardiovascular system are succeeded by those pertaining to the head and neck, gastrointestinal tract, extremities, and urinary system [6]. In order to promote a fuller understanding of the intricacies involved in the diagnosis, treatment, and moral decision-making surrounding this difficult genetic disorder, this case report aims to shed light on the complicated nature of Edwards syndrome.

The purpose of this case report is to add to the body of information already available about Edwards syndrome by shedding light on the complex features of this hereditary disorder. The goal of this case study is to improve our comprehension of the difficulties experienced by people who have trisomy 18 and the complex decision-making procedures that are a part of their care.

CASE HISTORY

One notable case originating from our institution involves a 22-year-old primigravida female who conceived spontaneously after a year of marriage with no familial history of chromosomal anomalies or prior miscarriages. The patient's newborn with 2.6 kg weight and 38 weeks of gestation, was admitted to the neonatal intensive care unit at the age of two hours due to mild respiratory distress, Oxygen therapy by nasal prongs and feeding were given during the stay and was discharged on breast feeding on the fourth day of life after sending investigation for chromosomal analysis. There were no complications till discharge. Immunization schedules and timely vaccination was explained to the parents. Chromosomal analysis revealed trisomy of chromosome 18 (Figure.1), accompanied by several distinctive features. These include congenital heart defects such as an atrial septal defect (ASD) and dysmorphic features such as malformed ears with flat pinnae (Figure.2), low-set ears (Figure.3), clenched hands with overlapping fingers, a flexed big toe and prominent heel, a shield chest or short and prominent sternum, and rocker bottom feet with a prominent calcaneum (Figure.4). Additionally, the patient exhibited ocular hypertelorism, microcephaly, hypotonia, a narrow bifrontal diameter, an open anterior fontanelle, a short nose, an asymmetric face, hypoplastic nails, flat feet, dorsiflexion of the great toe, a short neck with excessive skin folds and an umbilical hernia. On follow up developmental milestones appropriate for a one year of age, such as walking, beginning to speak simple words and engaging in social interactions. Weight and height were appropriate for one year of age and there was no any complications.

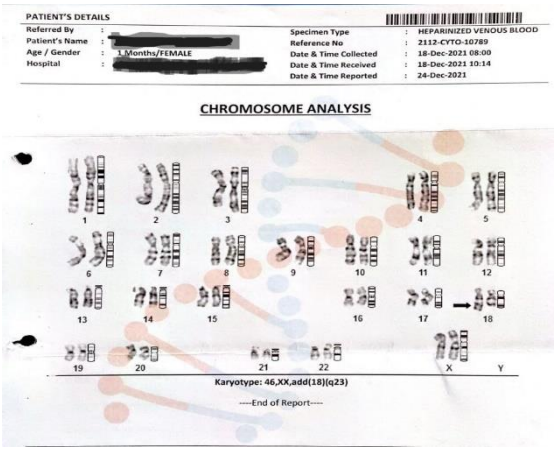

Figure-1: Trisomy of chromosome 18

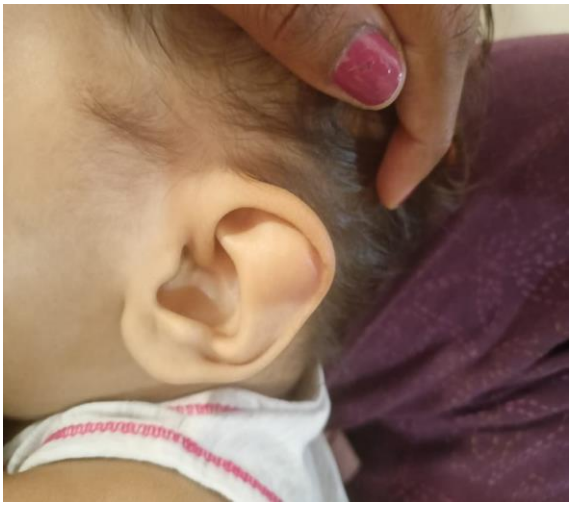

Figure-2: Malformed ears with flat pinnae

Figure-3: Low-set ears; Age: 1year

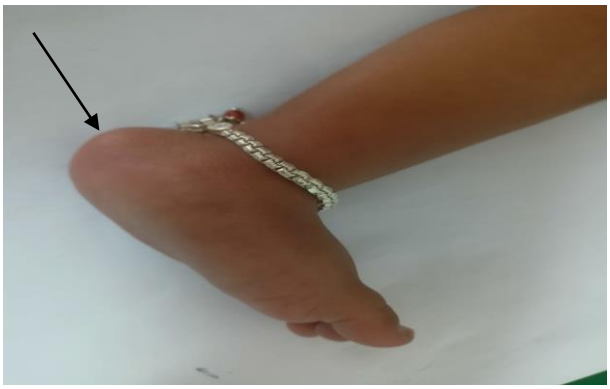

Figure-4: Rocker bottom feet with a prominent calcaneum; Age: 1year

DISCUSSION

Complexity of Genetics and Clinical difficulties linked to this chromosomal abnormality are highlighted by trisomy 18. Many physical and intellectual difficulties are caused by having an extra copy of chromosome 18, which emphasizes the necessity for a thorough understanding of the issue. It is impossible to exaggerate the significance of prenatal diagnosis in situations of Trisomy 18. Early detection is made possible by the use of non-invasive prenatal testing (NIPT) and confirmatory techniques such amniocentesis or chorionic villus sampling (CVS).[7] Consequently, this presents a chance for well-informed decision-making and permits suitable guidance to prospective parents concerning the possible difficulties and consequences linked to Trisomy 18. The case study highlights the value of a multidisciplinary care approach. Working together, genetic counselors, neonatologists, cardiologists, and other professionals can provide persons with Trisomy 18 with comprehensive and supportive treatment. By addressing the disorder's several medical and developmental elements, this integrated approach maximizes the standard of treatment received overall. The ethical conundrums that parents and medical professionals have when treating patients with severe genetic diseases are highlighted in this case study. The conversation that surrounds parental decision-making, including whether to continue or end the pregnancy, requires a careful balancing act between the family's values, medical knowledge, and the possible effects on the child's quality of life. Managing Trisomy 18 requires the use of palliative care. The affected person's and their family's overall quality of life must be prioritized, especially in light of the limited treatment options and seriousness of the related health problems. This raises questions about how medical professionals can support compassionate and dignified end-of-life care. This case study emphasizes the necessity of carrying out more investigation into the molecular processes that underlie Trisomy 18. Researching possible treatment options and learning more about the long-term consequences for those suffering from this illness may greatly advance medical understanding and enhance treatment plans.

Last but not least, the case report urges greater knowledge of Trisomy 18 in the community and among the general public. Improved public knowledge of the illness and assistance for impacted families can create a more compassionate and accepting community.

CONCLUSIONS

This case highlights the multisystem involvement of Edward syndrome (Trisomy 18), a rare genetic disorder confirmed by chromosome analysis. The child, now attempting to walk with a frame, exemplifies the varied clinical manifestations of the condition. Many cases are detected in utero through ultrasound and confirmed by amniocentesis. This instance adds evidence to the effectiveness of physiotherapy in managing Edward syndrome. Regular reviews and data collection of cytogenetically verified cases are crucial for understanding the relationship between genetic abnormalities and affected organ systems. The case underscores the importance of individualized care, from initial diagnosis to ongoing medical and developmental management, emphasizing the need for continuous research and awareness to improve the quality of life for those affected.

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