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A Case Report on Edward Syndrome

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Abstract

Background: Edward syndrome is commonly known as the trisomy 18 syndrome. It is a common chromosomal disorder due to the presence of an extra chromosome of 18. The feature of Edward syndrome involved in craniofacial, skeletal, cardiovascular, central nervous system and genitourinary malformations. Developmental delay is common in every child. Physiotherapy is recommended by several author to reduce the further progression of symptoms.

Case study: The case study was used to report this case and describe the physiotherapy management for child with Edward syndrome.

Discussion: A 6-year-old girl was diagnosed as Edward syndrome. The child is receiving physiotherapy treatment from June 2018. The treatment focused on improving active movement, posture, sitting balance, standing ability, respiratory status and others.

Conclusion: Edward syndrome is a rare autosomal chromosomal disorder. This case improved in developmental milestone. Edward syndrome can be identified by second trimester ultrasonography up to 97% cases in second and third trimester. Early provision of physiotherapy is very important to prevent further progression of symptoms and can helpful to reach developmental milestone as early as possible. This case adds our knowledge of physiotherapy management that had not been previously described.

Keywords: Edward syndrome; Physiotherapy

Introduction

Edward syndrome is a rare chromosomal disorder due to presence of extra chromosome on 18 that effect on multiple organs [1]. The live birth prevalence is from 1/3600 to 1/10000 [2]. Female and male ratio is approximately 3:1 [3]. The Edward Syndrome is commonly associated with cardiovascular problem including ventricular septal defect, atrial septal defect and patent ductus arteriosus. There is also found 90% of heart defect. The upper and lower extremities, urinary system, head and neck, gastrointestinal tract and genital system are affected [4]. Edwards Syndrome was first described in 1960 by Edwards et al. and Smith et al. by specific dysmorphic features [5].

The Edwards' syndrome phenotype consequences from full, mosaic or partial trisomy 18q. Full trisomy 18 is the most common form occurring in 94% of cases and every cell contains 3 full copies of chromosome 18. Mosaics can occur in 5% of cases in where some cells are normal with 46 chromosomes and others have the extra chromosome [6]. The clinical presentation of Edward syndrome is characterized by including: Low birth weight, craniofacial abnormalities: Low-set and malformed ears, micrognathia (small jaw), prominent occiput and dolichocephaly, small facial features for e.g., microphthalmia, microstomia, microcephaly, cleft lip and palate and/or narrow palate and coloboma of iris. The skeletal abnormalities: typical hand posture, clenched hands with index finger overriding middle finger and fifth finger overriding fourth finger, thumb aplasia, radial hypoplasia or aplasia, short sternum, hypoplastic nails, short, dorsiflexed hallux and prominent calcaneus (rocker bottom feet). The Congenital heart defects was found is about >90%, gastrointestinal abnormalities, urogenital abnormalities, neurological problems and Pulmonary hypoplasia [7].

Developmental delay is common with a profound degree of psychomotor and intellectual disability. In the most cases expressive language and independently walk are not achieved, but some older children can walk with the use of a walker. Though in children with Edward syndrome, developmental delay is markedly present so at every assessment of developmental progression is mandatory through standard developmental evaluation. There is also need for early referral to intervention programs and physical therapy is recommended [6].

Case Report

The child was 6-years-old, girl and came from urban area of Dhaka, Bangladesh. She has two brothers aged 18 and 15 years. Her parents both are graduate and lived in a single family. Their socioeconomic status was high. On the month of June 2018, the child came to CRP, Saver for better management according to Pediatrician advice. The physiotherapist used assessment to find out her problem. At first the therapist has done the subjective assessment. Before starting assessment, the therapist has taken the written and verbal consent from child's mother. The mother complaint of the child is not able to sit and walk independently (Figure 1).

Investigation

The impression of ECHO reported as Edward syndrome.

Inform consent

A written and verbal consent has given from child's mother before starting the formal assessment.

Assessment

The physiotherapist has done subjective and objective assessment to find out the problem. After finding problem therapist was proving following intervention in Table 1.

Results and Discussion

The aim of the study was to highlight physiotherapy for this child

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S/N	Impairments	Plan of care	Goal
1	Poor posture in sitting (supported)	Weight bearing practice at ischial tuberosity, practice upright sitting in cross-leg position and box	STG
2	Decreased active movement of UE and LE	ROM exercise	STG
3	Decreased spine range of motion (extension	Sustained extension in prone lying	STG
4	Problem in sitting balance	Sitting balance practice in physio ball and vestibular board	STG
5	Poor respiratory status	Breathing control exercise (abdominal & thoracic expansion)	STG
6	Poor position of foot	Prescribe AFO	STG
7	Gait problem	Standing balance practice in standing frame with AFO, stepping practice, gait training	To educate gait with walking frame (STG)
			To educate gait independently (LTG)
8	Poor higher function	Practice stair up and down, climbing, jumping if possible	LTG
9	Risk for developing secondary complications	Care giver education	STG+LTG
STG: Short Term Goal; LTG: Long Term Goal; ROM: Range of Motion Exercise; UE: Upper Extremity; LE: Lower Extremity; AFO: Ankle Foot Orthosis			

Table 1: Intervention according to impairments.

Figure 1: A 6-year girl diagnosed as Edward syndrome, and is trying to maintain her sitting balance in vestibular or balance board in physiotherapy session after receiving 6 months physiotherapy.

with Edward syndrome. After completing assessment therapist has provided above treatment according to plan of care. The time frame of short-term goal was 6 months and long term for 2 years. Still now this child is receiving physiotherapy. The mother of child is very concern about her child, and they are taking regular physiotherapy treatment 4 times in a week. Each session lasts for 45 minutes. The child is very cooperative with mother as well as therapist in therapy session. They are taking follow up from child neurologist timely for her other problem e.g., cardiac, gastrointestinal etc.

The purpose of this study was to report about Edward syndrome. In Edward syndrome, most common abnormalities were found in cardiovascular system, upper and lower extremities, urinary system, head and neck, gastrointestinal tract and genitals. The cardiovascular system associated with ventricular septal defect, atrial septal defect and patent ductus arteriosus. The calcaneovalgus defect, hip abduction and finger deformity are found in extremities. The microphthalmos, epicanthal fold and ocular hypertelorism were common in eye abnormalities in Edward syndrome. The low set ears are common in this syndrome. Micrognathia and short neck is also a common finding. Skull displays elongation defect with microcephaly. Mental retardation associated with both hyper and hypotonia is a common finding. In gastrointestinal tract, diaphragmatic hernia, umbilical hernia and pyloric stenosis are frequently seen. In urinary system, hydronephrosis, hydroureter with posterior urethral valve is commonly found. This current case presented with some of the above manifestations. This child now trying to walk with walking frame for short distance. A study found that a 4-year-old child with full trisomy 18 can walk independently. The diagnosis is confirmed by chromosome analysis. Many affected fetuses are detected by ultrasound scan during the second and third trimester of pregnancy and diagnosis can be confirmed antenatally by amniocentesis and chromosome analysis. This case adds some evidence to the literature about role of physiotherapy management for Edward syndrome.

Conclusion

Edward syndrome occurs due to meiotic nondisjunction with multisystem involvement. Most of cases are diagnosed prenatally by the basis of maternal age, detection of sonographic abnormalities during second and third trimester. So careful examination is very important to confirm diagnosis and it also help in identifying unexpected anomalies. There is very limited evidence on Edward syndrome. This case report may help to add in literature and helpful to compare with their known genetic disorder.

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