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Orthopaedics

PATTERN OF MUSCULOSKELETAL DEFECTS IN CHILDREN WITH TRISOMIES: A STUDY FROM NORTH KASHMIR

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ABSTRACT

Background: Multiple musculoskeletal abnormalities and concerns have been observed in chromosomal trisomies especially Downs syndrome. This can range from the affliction of spine in the form of atlanto-occipital and atlantoaxial instability to the involvement of feet as metatarsus primus varus. This observational study aims to highlight the importance of regular screening to allow timely detection and management of these anomalies.

Patients and methods: This was a community-based observational study, carried out in villages of North Kashmir. All the children with trisomy 21, 18, 13 based on phenotypic appearance were screened and confirmed by a paediatrician under RBSK (Rashtriya Bal Swasthya Karyakram).The patients were then examined by orthopaedic team for musculoskeletal abnormalities.The study was conducted over one year and six months from October 2018 to March 2020.

Results: Total 79 children with trisomies were examined, of which Downs Syndrome (DS) were 76 children, Edwards 2 children and Patau 1 child. The male:female ratio was 1.3:1. The median age of presentation was 3.9year for DS. Majority of children (92.40%) presented with delayed motor milestones, less common with pain, abnormal posture,deformityand limp. Feet abnormalities were the commonest, occurring in 86% of the cohort. A range of other musculoskeletal anomalies were observed involving knees (15%), spine and hips (12%), and knees (12%) with genu valgum (8.8%) and scoliosis (6.3%) occurring most frequently after pes planus (75.5%).

Conclusion : Children with trisomies especially DS are at increased risk of a number of potentially debilitating musculoskeletal problems. As the survival age of downs syndrome patients has increased the musculoskeleal anomalies can no longer be considered insignificant as well as their management. An annual musculoskeletal assessment for all children with DS could potentially enable early detection of problems, allowing for timely multidisciplinary team intervention and better clinical outcomes

KEYWORDS : Musculoskeletal anomalies - Trisomy - Downs Syndrome.

INTRODUCTION

Trisomy is characterized by the presence of 3 chromosomes instead of 2, of any particular chromosome. Trisomy 21 (Downs syndrome) is the most common trisomy with an incidence of 1 in 73 live births followed by trisomy 18 (Edward syndrome) 1 in 6000 births and Trisomy 13 (Patau)1 in 10000 births.[1]

Several musculoskeletal abnormalities and concerns have been observed in these chromosomal trisomies especially Downs Syndrome (DS). These include a range of anomalies like subluxation and dislocation of the cervical spine, hip and patella, scoliosis, metatarsus primus varus, and instability of the atlantooccipital and atlantoaxial region[2].Limb defects in the form of clinodactyly, polydactyly, rocker-bottom foot are frequently seen in Trisomy 13 and 18 [1].

Today the life expectancy Of Downs syndrome has increased to 55 years though most cases of trisomy 13 and trisomy 18 succumb to lethal congenital heart defects in infancy [3]. Our study aimed to determine musculoskeletal defects and orthopaedic morbidities in these patients aged up to 18 years, to highlight the importance of regular screening to allow timely detection and management of these anomalies.

PATIENTS AND METHODS

This was a community-based observational study, carried out in villages of Sopore Baramullah in North Kashmir. These villages fall in the catchment area of the Sub-District hospital of the place.

The study has been carried in Sopore,Baramullah, Jammu, and Kashmir, North India a Himalayan region of the state of Jammu and Kashmir, which is renowned for apple production and is also known as apple town. The Sopore town lies with geographical coordinates of 34030/N and 74047/ E. The total area of the study area is approximately estimated to be 320 square kms. The sub-district is home to about 2.2 lakh people, among them about 52% are males and about 48% are females. 100% of the whole population is from general caste, 0% are from schedule caste and 0% are schedule tribes. The child (aged under 6 years) population of Sopore Tehsil is 15%, among them 53% are boys and 47% are girls. There are about 34 thousand households in the sub-district and an average of 7 persons live in every family.

This cross-sectional survey was done along with the Rashtriya Bal Swasthya Karyakram (RBSK) mobile health team to identify birth defects, deficiencies, diseases, and developmental delays including disabilities in children

All the children with suspected chromosomal anomalies based on phenotypic appearance were screened by a Pediatrician under RBSK.To confirm the diagnosis karyotyping was done in these patients. Patients who were confirmed to have trisomy 21,18,13 were included in this study. The patients were then examined by an orthopedic team for musculoskeletal abnormalities. The patients were subjected to detailed clinical history and examination. A radiological investigation like X-ray, Computed tomography and MRI was done wherever indicated. The study duration was 18 months from October 2018 to March 2020.

Statistical analysis

The data obtained was accessed in SPSS software for necessary descriptive statistical analysis.

RESULTS

A total of 79 children with trisomies were assessed of which Downs Syndrome [DS] was 76, Edwards 2 children, and Patau 2. The median age of presentation of DS was 3.9 year for DS. Male: Female ratio was 1.3:1

Table 1: Demographic profile of children with known trisomies

	DOWNS SYNDROME n=76	EDWARD n=2	PATAU n=1
MEDIAN AGE AT SCREENING	3.9 year	2 month	1 month
MALE	n=44[56%]	n=1	n=1
FEMALE	n=32[42.1%]	n=1	

The majority of children n=73[92.40%] presented with delayed motor milestones less common was pain, abnormal posture, deformity, and limp. 31 [39.2%]were asymptomatic and were found to have a musculoskeletal abnormality on routine screening.

Table 2: Presenting complaint

DELAYED MOTOR MILESTONE	n=73[92.40%]
DEFORMITY	n=5[6.3%]
LIMP	n=3[3.79%]
PAIN	n=7[8.8%]

ABNORMAL POSTURE	n=5[6.3%]
AYSYMPTOMIC [screened under RBSK]	n=31[39.2%]

Table 3 shows the pattern of Musculoskeletal disorders. Feet abnormalities were commonest found in 68 [86%] children followed by a knee in 12 [13.18%].

Table 3: Pattern of musculoskeletal disorders among different anatomical regions

FEET	n=68[86.07%]
KNEE	n=12[15.18%]
SPINE AND HIP	n=10[12.65%]
HANDS /ARM	n=10[12.65%]

Table 4: The Pattern Of Musculoskeletal Disorders In Each Group

FEET	**n=68**
Pes planus	n=60[75.5%]
CTEV	n=2[2.53%]
Rocker bottom	n=2[2.53%
Hallus Valgus	n=4[5.06%]
KNEE	**n=12**
Genu valgum	n=7[8.8%]
Inflammatory arthritis	n=3[3.7%]
Patellar instability	n=2[2.53]
HANDS/ARM	**n=10**
Clenched hand	n=1[1.26%]
Polydactyl	n=2[2.5%]
Trigger digit	n=1[1.26%]
Overlapping fingers	n=2[2.5%]
Radial ray abnormality	n=1[1.26%]
Syndactyly	n=3[3.7%]
SPINE AND HIP	**n=10**
Cervical instability	n=2[2.53%]
Scoliosis	n=5[6.329%]
Hip instability	n=3[3.7%]

DISCUSSION

Over 18 months, 79 children with trisomies were examined, of which 76 were Downs syndrome (DS),2 had Edwards, and 1 had Patau. The median age of presentation was 3.9 years for DS. Two children with Edwards syndrome were2 months old and a child with Patau was one month old. Among children with DS 44[56%] were males and 32[42.1%] were females. All three children with Edwards and Patau syndrome were males. Male predominance was also seen in a large national study in Ireland by Foley et al and the median age of presentation was 8.9 years.[4]

The most common complaint for which children with DS were bought to a medical facility was motor developmental delay found in 73 children[92.40%]. Significant delay in walking was predominantly worrisome to parents. A delay in acquisition of motor milestones and lower levels of physical activity in children with DS have been observed[5]. Reduced physical activity contributes to the development of lower bone mass, obesity, and a failure to develop or maintain maximum possible muscle strength[6,7]. Inappropriately low expectations of physical activity and motor function from family, healthcare workers, and self feed into this cycle. Conversely, however, overattributing motor difficulties to low tone and hypermobility may lead to missed pathology and misdiagnoses.[8,9]

Less common presentations were deformity, pain and abnormal posture and gait.31[39.2%] children were incidentally found to have some form of musculoskeletal abnormality during screening commonest being pesplanus.

Feet abnormalities were found in 68 [86.06%]. Most common was pes planus found in 60 [75.5%]. CTEV in 2 [2.53%], rocker-bottom foot was seen in 2 children each with Edward and Patau syndrome. Hallus valgus was seen in 4 [5.06%] children.

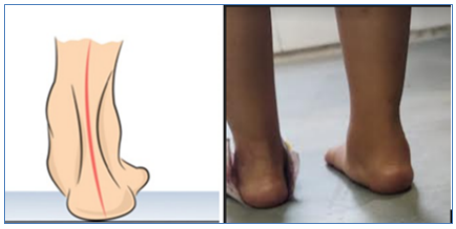

Figure 1

Pesplanus was seen in 91% of children in a study by Foleyet.al. [4].Young people with pes planus and incorrectfootwear are at risk of callus formation over pressure points, repetitive ligamentous injury, and development of bone spurs[10,11]. Inability of the calcaneus to come out of eversion leads to hindfoot valgus, resulting in multiple postural changes (Figure 1). This almost certainly contributes to the inability of many children with DS to sustain good strength when they stand or build good core musculature. Digital deformities also occur, such as hallux valgus.

Knee abnormalities were found in 12[15.1%] children. Genu valgum and genu varus were commonest found in 7[8.8%], Inflammatory arthritis in 2[3.7%], and Patellar instability in 2[2.53%]. The study by Foley et.al.[4]found a high incidence of inflammatory arthritis in 7.1%and Patellar instability in 1.5%. Genu varum/valgum may be added due to associated rickets and malnutrition seen in developing countries like ours.

Hand abnormalities were found in 10[12.6%] children. Syndactyly in 3 [3.7%], polydactyly and overlapping fingers each in 2[2.5%] children. Polydactyly and syndactyly were present in 0.5% and 0.3% respectively in the cohort by Foley et al.[4]

Abnormalities of spine and hip were seen in 10 children.Scoliosis in 5[6.3%], hip instability in 3[3.7%], and 2 [2.53%] children had cervical instability.C-spine instability is a well-described orthopedic condition associated with DS. It involves either the occiput-C1 level (atlantooccipital instability) or the C1–C2 level (atlantoaxial instability).

Incidence of cervical spine instability is significantly lower than previous studies that estimate 10%–27% of individuals with DS may have radiological findings of instability. However, most are asymptomatic, with only 1%–2% developing symptomatic instability.[11,12]

The low correlation between radiological findings and symptoms emphasizes the need for a high index of suspicion if symptoms develop.

CONCLUSION

Musculoskeletal anomalies in trisomies like downs syndrome are common and are usually underestimated. As the survival age of downs syndrome patients has increased the musculoskeletal anomalies can no longer be considered insignificant viz a viz their management. This observational study aims to highlight the importance of regular screening to allow timely detection and management of these anomalies. We advocate that such children should have an annual musculoskeletal assessment with an orthopaedician being an important member of the surveillance team.

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