

Knowledge and Awareness About Joubert's Syndrome Among Clinical Physiotherapy Practitioners

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ABSTRACT

Background: Joubert syndrome is a rare genetic condition characterized by abnormal brain development that includes the absence or underdevelopment of the cerebellar vermis (an area of the brain that controls balance and coordination) and a malformed brainstem. Clinical practitioners involve assessing, diagnosing, planning, implementing and evaluating treatment/interventions and care for the patients presenting with an undifferentiated diagnosis. Research has shown that physiotherapy clinical practitioners have satisfactory knowledge about JS.

Introduction: Joubert's syndrome is a rare autosomal recessive genetic disorder that affects the cerebellum, an area of the brain that controls balance and coordination. JS is estimated to affect between 1 in 80,000 and 1 in 100,000. Most of the signs and symptoms of JS appear very early in infancy with most children showing delays in gross motor and neurological features that include hypotonia, ataxia, developmental delay, intellectual disability, abnormal eye movements and neonatal breathing dysregulation.

Aim: This study was done to see the knowledge and awareness about JS among physiotherapy clinical practitioners.

Methodology: An observational study was conducted in Ahmedabad city. This study was approved by the Ethical Committee. Fifty-five clinical physiotherapists participated in this study. Physiotherapists completed a 16-item questionnaire of the knowledge and awareness about Joubert's syndrome.

Result: The result was carried out by using Microsoft Excel version 2016. Out of the 55-sample size, 75% of physiotherapists had knowledge and awareness about JS, which showed that there is good knowledge about JS.

Conclusion: This study concludes that there is good knowledge about Joubert's syndrome among clinical physiotherapists. However, better insight into this syndrome among clinical practitioners will give better opportunities for treatment.

Keywords: Joubert's Syndrome, Physical Therapy, Rehabilitation, Developmental support, Orthotics, Clinical Practitioner.

INTRODUCTION

Joubert's syndrome (JS) is an autosomal recessive genetic condition affecting the

cerebellum, the part of the brain responsible for balance and coordination. [1] It is a rare and intractable disease characterized by the

agenesis of the cerebellar vermis, episodic hyperpnea, hypotonia, abnormal eye movements, ataxia, developmental delay and psychomotor delay.^[2] The birth prevalence of JS is estimated to be between 1/80,000 and 1/100,000 live births, but JS may be under diagnosed and the true prevalence may be higher.^[3] Early hypotonia followed by marked ataxia and cognitive developmental delay. Typical facial features during childhood: -High rounded eyebrows, Broad nasal bridge, Mild Epicanthus, Anteverted nostrils, Triangular-shaped open mouth with tongue, frequently resting on the lower lip, Low-set and coarse ears. Characteristic neuro imaging findings as reported by Maria and colleagues with aplasia of the cerebellar vermis or marked hypoplasia showing only minimal connection between both cerebellar hemispheres with an umbrella shaped 4th ventricle, as well as stretched superior cerebellar peduncles with deep interpeduncular fossa leading to the “molar tooth sign”; of the mesencephalon. Supporting, but not mandatory features were the neonatal breathing pattern with tachypnea or apnea phases, eye motility problems (jerky movements, nystagmus, and oculomotor apraxia) and findings of retinal dystrophy.^[4] Joubert syndrome is an example of rare genetic disorder and knowledge among clinical physiotherapists about JS plays an important role in early identification, referral, care, coordination for individuals with physical and mental challenges, as well as chronic illness. A clinical practitioner involves assessing, diagnosing, planning, implementing and evaluating treatment/interventions and care for the patients presenting with an undifferentiated diagnosis.^[8]

MATERIALS & METHODS

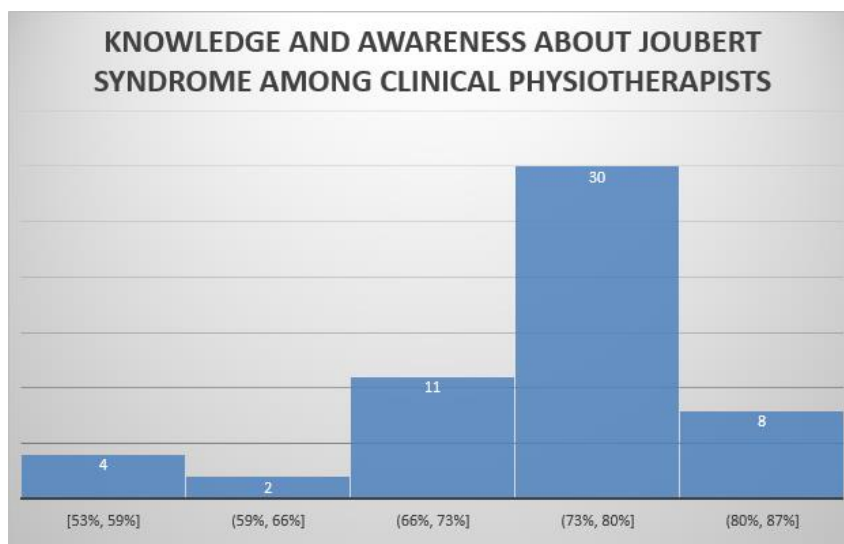
Questioner Method of Awareness about Joubert's Syndrome Scale was scored by Physiotherapy Clinicians. On a scale of 1 to 5 (1= Strongly Disagree, 2= Disagree, 3= Neutral, 4= Agree, 5= Strongly Agree). Subjects completed sixteen item questionnaires in which statements describes knowledge and awareness about the Joubert's syndrome. Age group 22 – 50 years with the working hour 7-8 hours in clinics or hospitals, only physiotherapists and those who were willing to participate in study were included and less than one year of experience, those who were not willing to participate and Non-medical practitioners (students) were excluded.

Sixty physiotherapy clinical practitioners were contacted and Questionnaires were sent. Information was gathered from fifty five physiotherapy clinical practitioners and five were dropped out. This study used cross-sectional observational research with random sampling. A Google Form was used to administer a survey. A range of various physiotherapy clinics were involved in this investigation.

RESULT

The result was carried out by using Microsoft Excel version 2016.

A total of 60 participants Out of the 55 participants responded and out of fifty five sample size, 75% of physiotherapists had knowledge and awareness about JS, which showed that there is good knowledge about JS. Most of the respondents were 30 years old and have more than 5 years of experience in the clinical practice.



DISCUSSION

In general, the findings revealed that there was a satisfactory level of knowledge and awareness among the physiotherapy clinical practitioners.

Dekair et al. did a study and reported a rehabilitation treatment program for a girl with JS focusing on improving the child's motor, language, cognitive, and social skills, including physical, occupational, and speech-language-hearing therapies; orthotic services improved her development but continued to be delayed for her age [5].

Regarding orthotic interventions, there have been reports of the prescription and use of a standard ankle-foot orthosis.

For hypotonia and ataxia, it is reasonable to improve the stability and limit the degrees of freedom of the lower limbs. Orthosis were considered in all three of our cases. Orthotic therapy should be considered to improve gross motor development and function in children with JSRD. [6]

In JS patients, while psychomotor developmental delay and emotional or behavioral problems vary, a positive correlation between age and equivalent age on cognitive and adaptive behavior tests has been reported, indicating that children with JSRD can continue to develop gradually and acquire new skills until adulthood. [7]

According to children's symptoms and developmental stages, appropriate rehabilitation approaches, including

physical, occupational, speech-language-hearing therapies and orthotic intervention, should be considered and provided to expand their activity and participation.

To raise more awareness about Joubert's syndrome among clinical physiotherapists and it is important to provide them with comprehensive information about the syndrome. This study included the details about the genetic cause, symptoms progression and availability of treatment options for Joubert's syndrome. Additionally sharing case studies and real-life examples can help physiotherapists to understand the impact of Joubert's syndrome on individuals and their families.

CONCLUSION

This study concludes that the knowledge and awareness of Joubert's Syndrome among clinical physiotherapists were satisfactory and better insight into this syndrome among clinical practitioners will give better opportunities for treatment. Awareness regarding Joubert's syndrome helps the physiotherapist individuals to plan a proper care and facilities. Future research with larger participation on physiotherapy students or other health care workers from different cities can be done.

Declaration by Authors

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