International Journal of Science and Research (IJSR)
ISSN: 2319-7064
SJIF (2022): 7.942

Evaluation of Motor Performance in a Child with Prader Willi Syndrome and its Influence on Mother's Quality of Life: Case Study

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Abstract: ***Background: Prader Willi Syndrome is a genetic disorder usually caused by deletion of a part of chromosome 15 passed down by the father. Prader Willi Syndrome is associated with hypotonia, distinct facial features, poor spinal reflexes, under developed genitals, delayed milestones, cognitive impairments, speech problems and behavioral issues. This affects their physical activity as well as motor performance. Due to long term complications, it negatively affects quality of life of a mother. The aim of the study was to evaluate motor performance of a child with PWS and to measure the quality of life of mother. Method: In this study Peabody developmental motor scale -2: gross motor scale and Early clinical assessment of balance were used for the evaluation of motor performance of a child; and SF-12 and Parental stress scale were used to examine the Quality of life and magnitude of the stress level of a mother suffering from PWS. Result: Conclusion: There is significant delay in gross motor function of a child with moderate delay in fine motor function. There is significant balance affection in a child with PWS due to motor involvement. Parental stress is at moderate level with affection of role of limitation due to physical health in SF-12.***

Keywords: Prader Willi Syndrome, Early Clinical Assessment of Balance, Peabody Developmental Motor Scale 2, SF-12, Parental Stress Scale Questionnaire.

1. Introduction

Prader Willi syndrome is a multisystemic complex genetic disorder caused by a lack of expression of genes on paternally inherited chromosome 15q11.2-q13 region. There are three main subtypes of Prader Willi syndrome(PWS) maternal uniparental disomy, parental deletion, and imprinting defect. Prader Willi syndrome was first described by Prader et al and now it is termed a genetic disorder.[1]

Prader Willi syndrome is a rare genetic disorder, it affects both population males and females equally. Incidence of disorder is 1/10000 to 1/30000. The prevalence of the condition is 10000 to 20000 in the United States. This condition affects hypothalamic functions, including multiple systems throughout the body.[2]

There are many causes of PWS, most commonly cause paternal deletions, imprinting centre (microdeletion of 15q chromosome) defect, and maternal disomy (15) (both contributed by the mother).

35% of cases are caused by maternal disomy and 5% of cases are affected by imprinting center defect. Other causes of Prader Willi Syndrome are hearing loss, cardiac abnormalities, seizures, or metabolic defects due to several recessive genes. Risk factors of this condition due to advanced maternal age, growth hormone deficiency, and hypothyroidism.[3]

Clinical symptoms of Prader Willi syndrome are varies depending upon age group. During infancy, there will increase the risk of seizure, periodic breathing with apneas, and instability in temperature. Temperature below 90 degrees was noted in adolescents and adults. Craniofacial abnormalities and neuromuscular weakness are commonly seen.[4]

Prader Willi syndrome has also been influenced by other factors also like the lifestyle of parents, relationships with siblings, and social impact. Early diagnostic manifestations of paternal and maternal can be beneficial to prevent a child from Prader Willi syndrome.[5] Physical, emotional, social, cognitive, communication, and activity of daily living all of these are affected by the different age groups of children suffering from Prader Willi Syndrome. Even caregivers are also affected physically and mentally in looking after the child. The most common factor that affects them is Worry for them.[6]

In spite of all these factors which are affected in Prader Willi syndrome, cognitive impairment is one of the impairments in them. Children have lower cortical complexity in the frontal, temporal, and parietal lobe.[7]Impairment in the motor, emotional and cortical influence of the child's parent as well as a sibling. Due to this, they have less participation in society, which can affect their quality of life.[8]

In the present study, the following scales are used to evaluate the quality of life of the mother, SF-12 (reliability =0.80) is used, and for stress level Parent Stress Scale questionnaire(reliability= is used. To evaluate gross motor and fine motor function, the Pea Body scale is used(test-retest reliability = 0.99). To examine balance Early Clinical Assessment of Balance version-2 is used.

Prader Willi syndrome not only affects the child but also influence parents' life, and can be a stress factor for them. To relate the level of stress and affection to their quality of life, a study is need to be done.

Need for the study

Prader Willi syndrome is a rare genetical condition which involves major systems of our body like motor function, cognition and behavior. As the condition is less explored and need to study more with research literature, our study may help to conclude about the symptoms and average delay in milestones of a child.

Aim of the study

Aim of the study was to evaluate the gross motor and fine motor delay of a child with PWS and to examine the quality of life of a mother.

2. Method and Materials

A case study on a female child who was diagnosed with PWS was included in our study. We assessed gross motor and fine motor functions, balance of a child and quality of life of a mother.

Outcome measure

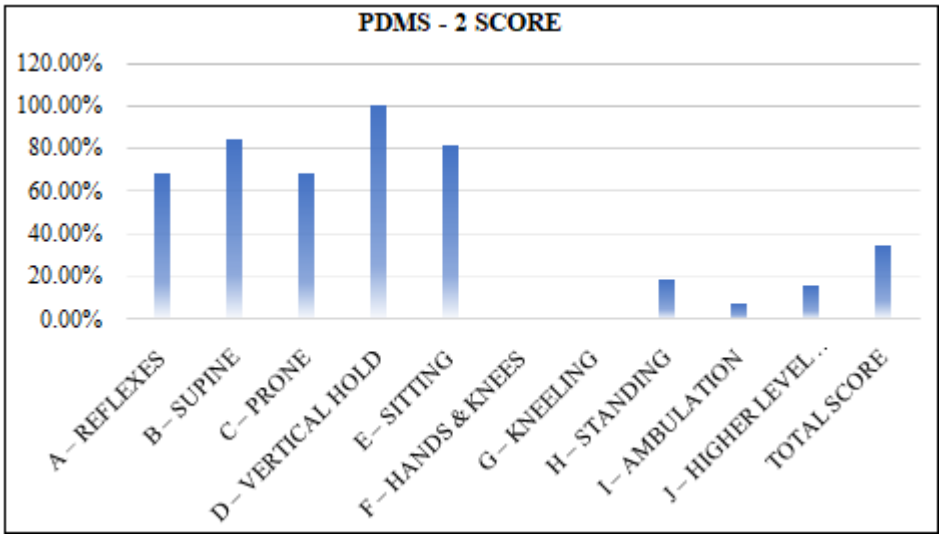
1) Peabody developmental motor scale 2 (PDMS)
2) Early clinical assessment of balance (ECAB)
3) SF – 12
4) Parental stress scale

3. Results

a) Peabody developmental motor scale – 2

Subscale	Score	Percentage of Performance
A – REFLEXES	11/16	68.75%
B – SUPINE	22/26	84.61%
C – PRONE	11/16	68.75%
D – VERTICAL HOLD	8/8	100%
E – SITTING	26/32	81.32%
F – HANDS & KNEES	0/8	0%
G – KNEELING	0/2	0%
H – STANDING	6/32	18.32%
I – AMBULATION	4/58	6.89%
J –HIGHER LEVEL ACTIVITIES	17/108	15.74%
TOTAL SCORE	105/306	34.41%

Total score of the child is suggestive of severe gross motor delay in the milestones of the child which is matched with the peer group.

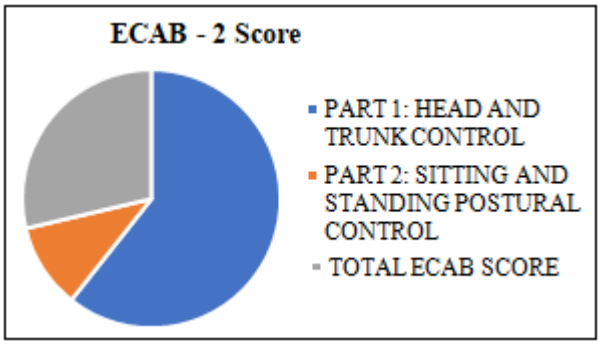


b) Parental stress scale:

Total score of the scale is 59/90 which further interprets moderate level of stress in parent of the case.

c) Early clinical assessment of balance – 2:

Part	Score	Percentage of Performance
Part 1: Head and Trunk Control	32/36	88.88%
Part 2: Sitting and Standing Postural Control	10/64	15.62%
Total ECAB Score	42/100	42%



d) SF – 12:

Component	Score	Percentage of Performance
PCS-12	49.60/56.57	87.67%
MCS-12	60.28/60.75	99.22%

4. Discussion

The purpose of the study is to determine the affection of child suffering from Prader Willi Syndrome to a mother's quality of life and stress. A child suffered from Prader Willi syndrome requires a lot of affection from parents and siblings too. The present study showed that physical impairment of child and retard in growth develop parent stress also affects the quality of life. In the present study age of the child is 2 years, diagnosed with PWS.

Her physical therapy treatment included sit-to-stand exercise, gait training with a walker, proprioceptor training, and balance training. Her mother had a moderate level of stress, from the parental stress scale she scored 59/60. In the study, Linda et al[9] showed that growth hormone had an effect on gross motor and fine motor development. To assess motor impairment pea body development scale is used in the study. Results suggested severe gross motor delay development in milestones of the child. To assess the quality of life, SF-12 is used in the present study, PCS-12 score is 49.60 & MCS-12 score is 60.28 which showed there is more impact on physical than mental components.

Kathleen et al[10] in their study used the Peds QL scale to assess the quality of life they had compared with children suffering from PWS and non – PWS. In their study parents have more affection in the emotional subscale and less in social whereas in the present study physical component is more affected.

In the present study, for balance assessment, early clinical assessment of balance is used. Results showed that standing and sitting postural control is more affected than head and trunk control. The total ECAB score was 42/100. Reus et al[11] used muscle thickness assessed by ultrasound and infant muscle strength meter to evaluate motor performance and strength which is not measured in the present study.

The current study showed due to children suffering from Prader Willi syndrome, parents are having more stress and it affects their quality of life. The physical activity of parents is more affected rather than mental and social.

5. Conclusion

There is significant delay in gross motor function of a child with moderate delay in fine motor function. There is significant balance affection in a child with PWS due to motor involvement. Parental stress is at moderate level with affection of role of limitation due to physical health in SF-12.

Acknowledgement
First, we would like to thank the almighty lord for his blessings. We would like take opportunity to thank our parents for their constant support, encouragement and motivation.

Conflicts of interest
Nil

Funding
None

Ethical clearance
It was given by ethical committee

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