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JOUBERT SYNDROME

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ABSTRACT

Joubert syndrome (JS) is an unusual neurodevelopmental disorder. Its trait of hypotonia, ataxia, and delayed developmental milestones. It affects multi system such as respiratory, neurological, renal, ocular, liver, limb. Many genes involved, problems occur in the site of cilium where it joins with the plasma membrane[1]. It is also called ciliopathy. Patient had feeding problems, hearing and vision problems. Confirmed the diagnosis by molar tooth sign on magnetic resonance imaging and Genetic testing. The patient will be managed by supportive treatment[2] with the help of multi team members. Many patients need rehabilitation and genetic counselling.

KEYWORDS

Joubert Syndrome, Neurodevelopmental disorder, Genetic testing, Cilia and Molar tooth sign.

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INTRODUCTION

Joubert syndrome (JS) is an uncommon genetic neurodevelopmental disorder. It characterized by abnormal development of the cerebellum and brainstem. Problems occur in coordination, breathing, eye movements and development. Many genes involved (e.g., AHI1, CEP290, TMEM67, *CPLANE1*). Mutations occur in these eight ciliary/basal body genes such as INPP5E, AHI1, NPHP1, CEP290, TMEM67/MKS3, RPGRIP1L, ARL13B and CC2D2A[3].

Cause

Usually autosomal recessive inheritance

Caused by mutations in genes affecting primary cilia (classified as a ciliopathy)

Clinical Features

Neurological

Hypotonia (floppy infant)

Ataxia (poor balance, unsteady gait)

Developmental delay

Intellectual disability (variable)

Respiratory

Abnormal breathing pattern in infancy (episodes of apnea and hyperpnea)

Ocular

Abnormal eye movements

Oculomotor apraxia

Nystagmus

Visual impairment

Facial Features

Broad forehead

Arched eyebrows

Ptosis

Open mouth appearance

Retinal dystrophy[4]

Associated System Involvement (may be present)

Kidneys: nephronophthisis → renal failure

Liver: congenital hepatic fibrosis

Eyes: retinal dystrophy

Limbs: polydactyly, syndactyly[5]

Mouth: Cleft lip and palate[5], soft-tissue masses on the dorsum of the tongue[6]

Diagnosis

Prenatal

DNA test, Fetal MRI[7]

Newborn

MRI brain – Molar tooth sign, Bat wing sign[8], cerebellar vermis hypoplasia (CVH), deepened interpeduncular fossa and thick, elongated superior cerebellar peduncles[7].

Genetic testing - Whole exome sequencing (WES) analysis, Optical genome mapping (OGM)[9] and sanger sequencing[10]

Clinical assessment: The Gesell Developmental Scale is using to evaluate the intelligence level[8]

Video electroencephalogram (VEEG) -diffuse slow waves with overlapping sharp waves during wakefulness and sleep

Management

No cure

Supportive and multidisciplinary care

Physiotherapy and occupational therapy

Speech therapy

Regular monitoring of kidney, liver, and vision

Genetic counseling for parent

Management of breathing problems

Altered respiratory control requiring mechanical ventilation and/or tracheostomy

Management of feeding difficulties

For children with significant feeding difficulties related to hypotonia or dyscoordinated oromotor function, nasogastric feeding tubes or gastrostomy placement have been necessary to prevent aspiration and provide adequate caloric intake[7].

Endocrine disorders may require hormone replacement.

Nursing theory application

Roy's Adaptation Model

Concept: Child and family continuously adapt to internal and external changes.

Outcome: Improved adaptation; stable physical health; emotionally supported parents.

Prognosis

Highly variable

Depends on severity and involvement of organs

Early intervention improves quality of life.

Nursing care plan:

Ineffective Breathing Pattern related to brainstem dysfunction

Goal: Maintain normal RR and SpO_2; baby free from apnea

Interventions:

Monitor RR, rhythm, SpO_2 continuously

Position head elevated

Provide oxygen if needed

Use apnea monitor

Gentle stimulation during apnea

Risk for Aspiration related to poor suck–swallow coordination

Goal: Baby feeds safely without choking/aspiration

Interventions
Assess swallowing ability
Upright feeding position
Slow, small frequent feeds
NG tube if unsafe feeding
Burp frequently
Imbalanced Nutrition: Less than body requirement related to feeding difficulty
Goal: Adequate nutrition and weight gain
Interventions
Daily weight monitoring
Dietitian referral
Thickened feeds/NG feeding
Monitor hydration and I/O
Encourage breastfeeding
Delayed Growth and Development related to neurological impairment
Goal: Promote optimal developmental progress
Interventions
Assess milestones regularly
Physiotherapy, occupational therapy
Positioning and passive exercises
Sensory and visual stimulation
Parent teaching
Risk for Infection related to hospitalization and weak immunity
Goal: Baby remains infection free

Interventions
Strict hand hygiene
Aseptic technique
Immunization updates
Monitor temperature
Parental Anxiety / Caregiver Role Strain related to chronic condition
Goal: Parents understand condition and cope well
Interventions
Explain disease clearly
Teach home care techniques
Emotional support and counselling
Support group referral

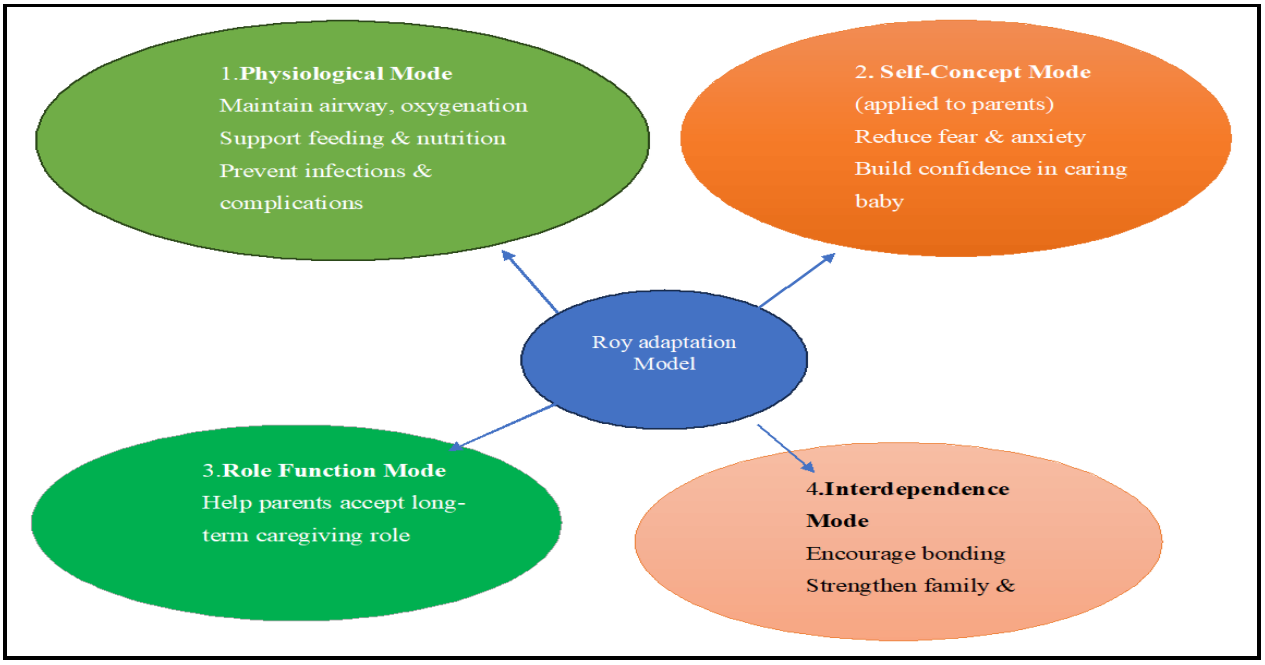

Figure No.1: Roy's Adaptation Model

CONCLUSION

Nursing care for children with Joubert syndrome plays a vital role in improving quality of life, preventing complications and supporting families through long-term care needs. Through early identification, continuous monitoring of respiratory status, developmental progress and associated organ involvement, nurses help ensure timely interventions and coordinated multidisciplinary management.

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CONFLICT OF INTEREST

We declare that we have no conflict of interest.

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