WORLD JOURNAL OF PHARMACEUTICAL RESEARCH
SJIF Impact Factor 8.074
Volume 7, Issue 14, 939-945. Case Study ISSN 2277– 7105

JOUBERT SYNDROME: A CASE STUDY

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Article Received on
01 June 2018,

Revised on 21 June 2018,
Accepted on 11 July 2018,

DOI: 10.20959/wjpr201814-12942

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ABSTRACT

Joubert syndrome is an autosomal recessive disorder associated with agenesis of cerebellar vermis of the brain that controls balance and coordination. It is characterized by cerebellar vermis hypoplasia, hypotonia, and one of the two conditions: abnormal breathing pattern and or abnormal eye movement. Here we present a case study of 4.5 years old male diagnosed with joubert syndrome having major complaints of delayed milestones, hypotonia and oculomotor apraxia. At present no treatment is available on joubert syndrome in modern medical science. The assessment was monitored through GMFCS score, which is not only flaw free but gives exact improvement status along with Development quotient (DQ) which was also assessed to ascertain whether the velocity of development is achieved or not. Study reveals the management of Joubert syndrome based on *Ayurvedic* principles which proved to be very beneficial in this case.

KEYWORDS: Joubert syndrome, autosomal recessive disorder, oculomotor apraxia, GMFCS, DQ.

INTRODUCTION

Joubert syndrome is first described by Joubert in 1969.[1] The incidence of Joubert syndrome is between 1/80000 and 1/100,000.[2] Joubert syndrome is very rare disorder associated with absence or underdevelopment of cerebellar vermis (a part of the brain that controls balance and coordination) and malformed brain stem (connection between the brain and spinal cord).[3] It is a multisystem disorder which mainly affects nervous, urogenital, ocular, and gastrointestinal system in varying proportions. The onset of disease occurs during the

gestation period when the brain development takes place. No definite treatment available for Joubert syndrome. Available options of treatment are physiotherapy, occupational therapy etc.

According to *Ayurveda* proper *Sharira cheshta* is *karma* of *vata*.[4] Vitiated *Vata* causes abnormality in tone, posture and movement of the body. The movement of eyes,[5] and speech,[6] is also controlled by *Vata* only. Hence treatment protocol was planned aiming to normalize *Vata* and improvement of quality of life of the patient. *Snehana*, *Swedana* and *Basti karma* are the main pillars of *Vata chikitsa*,[7] Studies have shown that mean age of death in Joubert syndrome is around 7.2 years and it‟s mainly due to respiratory failure.[8] Hence, *Rasayan chikitsa*,[9] was mainly included in treatment protocol to maintain the improvement, to improve immunity and to prevent crippling of child. Squint, nystagmus and delayed language are the associated complaints hence *Sthanik chikitsa* like *Ashchotan* and *Jiva pratisaran* are also included in the study.

In Joubert syndrome multiple systems of body are involved, hence multidisciplinary management was planned including both internal medications and *Panchakarma chikitsa.*

CASE DETAILS

A 4.5 years old male child weighing 15 kg was brought to *Kaumarbhritya* OPD by his parents with the chief complaints of global delayed milestones, nystagmus and ataxia (balance problem). The boy was not able to stand without support, could speak only 2 bisyllables, and was already achived his sitting milestones at the age of 2.5 years. The boy used to get easily irritable and was bit hyperactive. Social milestone was well achieved as compared to motor, language or cognitive milestones.

By taking detail history from the mother and reviewing the medical file following points were observed: the child was a product of a cesarean section at full term (indication was CPD). There was no h/o NICU stay. From the age of 1.5 years the baby had recurrent episodes of breath holding spells which was lasts in PICU admission for 8 days. Regarding the family history; patient had no siblings, parents had second degree consanguineous marriage. No similar presentation in any of family members was found. The prenatal history was insignificant.

Physical examination revealed the following

Easily irritable child, mild generalized hypotonia, delayed developmental milestones with gait ataxia, frontal bossing, small low set ears, deep seated eyes, strabismus, Rt. side narrow palpebral fissure, hypertelorism, depressed nasal bridge, broad root of nose, bullous nasal tip, retrognathia, inverted „V" shaped upper lip, high arched palate etc.

Ocular examination

- Patient could not follow moving objects with ease.
- Horizontal nystagmoid movement of eyes.
- Signs of oculomotor apraxia noted.
- No eye movements noticed in vertical meridian. Squinting was present.

Diagnosis

- Clinically on the basis of signs and symptoms.
- Also investigations s/o joubert syndrome.

Investigations

- **Neurosonogram:** Both lateral ventricles are minimally dilated. Suspicious abnormality seen in cerebellum.
- **CT scan:** Widening of biparietal and bifrontal CSF spaces. Most probably benign enlargement of subarachnoid spaces. No focal brain lesions, central midline structures, no hemorrhages or collections seen.
- **Brain MRI:** Vermian hypoplasia with midbrain having "molar tooth" appearance. This is consistent with Joubert syndrome.

Prevention

- Physiotherapy for the duration of 6 months

Ayurvedic Protocol Used

Sr. no.	Treatment	Content	Duration
1	*Abhyanga* (Oleation)	*Tila taila*	Daily 15 min for 6 months
2	*Swedana* (Sudation)	*Shashtishalik pinda sweda* with *shatavari, bala* and *ashwagandha sidhha ksheer*	Daily for 40 min for 6 months
3	*Basti (Enema of medicated decoction or oil or any liquid)*	*Baladi basti* (*bala, ashwagandha, shatavari, musta, guduchi,yashtimadhu*) *Anuvasan* – 30 ml *Tilatail*, *Niruha*-200 ml *Kwath*.	(*Karma basti krama*) 3 cycles
4	*Abhyantar chikitsa*	*Kavachbeej choorna* 5g in 1 cup of milk	(BD) for 6 months
5	*Rasayan chikitsa*	*Kumarkalyan rasa* (100mg), *Vatagajankush* (250mg), *Vishatinduk vati* (125mg), *Samvardhan ghrita* (5ml)	(BD) for 3 months
6	*Ashchotan*	*Shatavari ghrita*	2 drops daily
7	*Jiva pratisaran*	*Vacha* and *akkarkara churna* (250mg)	BD

Assessment Criteria and Observations

Assessment was done on basis of Developmental quotient (DQ) which was taken under four headings.

- Gross motor DQ
- Fine motor DQ
- Language DQ
- Social DQ
- Total DQ and GMFCS scoring also on clinical examinations.

Table 1: Observations.

DQ	Before treatment	After 1.5 months	After 3 months	After 4.5 months	After 6 months	After 8 months
Grossmotor	19.29	20.8	22.03	39.34	50	52.17
Fine motor	21	41.37	45.76	59.01	67.74	86.95
Language	15.7	20.8	23.72	29.50	33.87	33.87
Social	63.1	63.1	63.01	78.68	80.64	78.26
Total	29.77	36.19	38.13	51.63	58.06	62.81
GMFCS score	45.6	55.33	60.21	73.71	77.03	87.96

Probable Mode of Action of *Ayurvedic* Protocol

Ayurvedic regimen normalizes the vitiated *Vata*. It also gives strength to muscles and improves eye coordination and speech development. *Snehana* and *Swedana* provide

cutaneous manipulation. It is one of the important procedures for mitigating *Vata*.[10] Cutaneous stimulation results in increased circulation which enhances transdermal drug absorption which in turn helps to normalize the muscle tone,[11] of the patient. *Baladi basti* contains six drugs (*bala*[12] *ashwagandha*,[13] shatavari,[14] musta,[15] guduchi,[16] *yashtimadhu*)[17] having predominant *Vatahara* and *Rasayana* properties. It is a *Niruha basti* which causes *Shodhana* as well as increases *Bala*, also normalizes the bowel and bladder control. It helps to prevent the tendency of chronic constipation.

Rasayana,[19] treatment is the blessing of *Ayurvedic* science to medical science. It is useful for enhancement of quality of life, longevity, intellect and physical strength. Use of *Kumarkalyan rasa*,[20] *Vatagajankush rasa*,[21] *Vishatinduk vati*,[22] along with *Samwardhan ghrita* leads to nourishment of vital tissues, improvement in speech, intellect and prevents crippling hence results in improving the quality of life. *Ashchotan* and *jiva pratisaran* work as a *sthanik chikitsa* according to major symptoms of patient.

RESULTS AND DISCUSSION

In this patient social milestone achievement was already better than other developmental milestones. Gross and fine motor milestones improvement was better than language improvement. Marked hypotonia was improved within 2 months of treatment, and at present is having almost normal muscle tone. He has achieved controlled walking without support, can take turn without losing balance. Nystagmus was controlled after 1 month of treatment, after 6 months of treatment he became able to follow an object without any nystagmus. Mild squint is still present. He has achieved his bladder and bowel control after completion of 2 cycles of *Basti chikitsa*. Fine motor development is achieved almost normal as that of the 5 years of child and the velocity was maintained constant. There was no marked improvement in speech even after 3 months of treatment but from that onwards the child started improving his vocabulary and present status is that he can use 8-10 word with meaning. Child is not able to run yet and management is still going aiming at further improvement.

ACKNOWLEDGEMENT

Author is thankful to his guide and co-guides for their encouragement towards research work.

CONCLUSION

This *ayurvedic* treatment protocol is proved to be beneficial in above case of Joubert syndrome mainly in the domain of gross motor and fine motor.

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