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Case Report

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AN AYURVEDIC APPROACH ON MUCOLIPIDOSIS TYPE II – A CASE REPORT

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ABSTRACT

Mucolipidosis II, also known as I- cell disease, is a rare disease which is a progressively debilitating disorder that affects many parts of the body. Although its exact prevalence is unknown, it has been estimated that 1 in 100,000 to 4, 00,000 individuals worldwide are affected. Themutation of GNPTAB gene is found to be the cause of Mucolipidosis II. Here an effort is made to explain the *Vatahara* line of treatmenton a case of 6 year old childpresenting with weakness of the limbs especially the lower limb along with, delayed milestones and mental retardation with facial dysmorphism, by considering the concept of *Beejabhagaavayava Dusti* mentioned by Chakrapani.

KEYWORDS: Mucolipidosis type II, *Beejabhagaavayava Dusti, Vatahara* treatment.

INTRODUCTION

I-Cell disease is a rare inherited metabolic disorder characterized by coarse facial features, skeletal abnormalities and mental retardation. This disorder belongs to a group of diseases known as lysosomal storage disorders. Lysosomes are particles bound in membranes within cells that break down certain fats and carbohydrates. Multiple enzyme deficiencies associated with I-cell disease lead to the accumulation of certain fatty substances (mucolipids) and certain complex carbohydrates (mucopolysaccharides) within the cells of many tissues of the body. I-cell disease is caused by a mutation in the GNPTAB gene that leads to a deficiency in the enzyme UDP-N-acetylglucoseamine-1-phosphotransferase. I-cell disease is inherited as an autosomal recessive genetic trait. Children with I-cell disease typically have severe delays in the development of gross and fine motor skills, hearing loss, lack of muscle tone (hypotonia), and varying degrees of mental retardation. Other symptoms of I-cell disease may include frequent respiratory infections, constipation and/or diarrhea, overgrowth of gum tissue (gingival hyperplasia), and/or joint stiffness or joints that are "frozen" in place (contractures).

I-cell disease (Mucolipidosis II) is caused by a mutation in the GNPTAB gene that is located on the long arm of chromosome 4 (4q21-q23). The GNPTAB gene mutation leads to a deficiency in the enzyme UDP-N-acetylglucoseamine-1-phosphotransferase that is involved in the synthesis of mannose-6-phosphate, resulting in decreased intracellular levels of lysosomal enzymes and increased levels in blood serum and body fluid. The symptoms of I-cell disease develop due to deficiencies of a variety of lysosomal enzymes in the cells of the body causing an abnormal accumulation of certain fatty substances (mucolipids) and certain complex carbohydrates (mucopolysaccharides) within the cells of many tissues of the body.

Ancient *Ayurveda* scholars were very much curious to know causes of these various anomalies. *Vedas, Brahmana Granthas, Upanishads, Smrutis* and most of *Ayurvedic* texts have reference, concerned to these problems. As per *Charaka Samhita, Punnarvasu Atreya* says that abnormalities of *Bija* (sperm and ova), *Atma Karma, Asaya Dosa* (uterus), *Kala* influences the growing foetus. *Susruta* has described that any disease produced by any one or combined with following seven causes like *Adibalapravrutta, Kalabalapravrutta, Sanghatabala, Daivabala* and *Swabhavabalapravrutta*. Out of these, congenital malformations are caused by A*adibalapravrtta and Janmabalapravrutta* factors. Abnormalities in *Beeja* are responsible for various malformatios in the fetus. These malformation depends on the *Beeja*. The anomaly of the body depends upon the affected portion of *Beeja*. *Charaka Samhita* says that an organic abnormality cannot occur without abnormality of corresponding portion of *Beeja*, *Chakrapani* has explained various terms like *Beeja, Beejabhagavayava*. This *Beejabhagaavayava* are concerned with the development of various different organs.

The defect in the *Beejabhagaavayava* is responsible for the congenital anomalies and the treatment of these condition are considered to be *Yapya*.

Way of Evolution
A child of 6 years old was presented with complaint of weakness of limbs especially on the lower limbs, delayed development, unable to speak, dribbling of saliva and intermittent banging of head with mental retardation. The child was delivered through emergency LSCS (due to arrested descent). Baby cried after the resuscitation and was referred to higher center for further management. After 8 hrs the child had subtle seizure for which the treatment was under went and the child was kept under observation. Later, during the 48th day the child again developed tonic seizure along with the respiratory distress for which the child underwent treatment. Later parents noticed global delay in milestones as well as the mental retardationfor which they were undergoing treatment and did not find much improvement. So got admitted in S.D.M college of Ayurveda & Hospital.

Birth History
Antenatal
- A G3P2A1L2 mother who was residing in Endosulphan affected area in Kasaragod.
- One child got aborted due to Hydrocephalus.
- The mother had under gone routine antenatal history.

Natal
- Emergency LSCS due to arrested descent.
- Cried after resuscitation.
- Birth weight – 3.9kg.

Post Natal
- Subtle seizure developed after 8 hrs.
- Neonatal encephalopathy.

History of Past Illness
H/O Tonic seizure.

Treatment History
- SypGardenal - 3ml BD
- SypEptoin – 2ml BD
- SypShelcal – 5ml BD
- Under went Physiotherapy

Family History
No family history and consanguinity found.

Immunization History
Appropriate for age.

Personal Histor
Aharaja
Patient was totally dependent for food intake, appetite was poor. Diet was dominant in *Madhura Rrasa* (sweet diet).

Vihāraja
Nature of activity was always assisted. Sleep was disturbed (2–3 h/day, 6–7 h/night). Repeated banging of the head and drooling from the mouth was present.

Examination
Vitals were normal. Cardiovascular system, and per abdomen examinations had shown no deformity. *Prakṛti* (constitution) was *Vātādhikakapha*.

Respiratory system: on auscultation showed presence of ronchi.

Aṣṭa Vidha Parīkṣā
Nāḍi (pulse) was *Vātādhika Tridoṣaja*. There was no complaint with regard to *Mūtra* (urine). Frequency and color were normal. *Mala* (stool) was constipated, once in 2–3 dayṣ Bowel control was not yet achieved (which should have been achieved by 1.5 years of age). *Jihvā* (Tongue) was *Sāma* (coated suggestive of improper digestion), macroglossia, *Śabda* (speech) mumbling. *Sparśa* (touch) was hard and dry. *Dṛk* (eyes) flat eye. *Akṛti* (appearance) was normal nourished with coarse facial dysmorphism.

Motor system examination
Patient was diagnosed to have the stiffness of joint was present along with deformity in the ankle joint. Muscle power could not be elicited because patient was unable to follow the command. Hyperreflexia was present in the lower limbs. Sensory system was intact, and no abnormality found. Cranial nerve examination could not be done because of severely handicapped physical and mental state of the patient. Meningeal signs were not present.

Differential Diagnosis
Spastic CP, Demyelinating (degenerative) disease of central nervous system (CNS), Mucopolysacchridosis.

Final Diagnosis
Mucolipidosis type II.

Treatment Protocol
- Udvartana with Triphala for 2 days
- Abhyanga with Shishuposhakataila and Godhumapinda Sweda for 5 days.
- Matrabasti with Dhanvantaraghrutha for 7days.
- Shirolepa with Amalaki, Vacha and Yashti for 7 days

CONCLUSION
The thorough analysis of the case shows that there occurs a *Vatadusthi* in the *Beeja* which in turn lead to the *Beejaavayavadusthi* which is causing the *Dusthi* of the *Rasa* and *Rakta Dhatu*thereby responsible for the mental retardation and the weakness of the limbs and delayed development. The main *Srotas* involved can be said as *Pranavahasrotas, Annavahasrotas* and *Manovahasrotas* which are responsible for the symptoms. By considering

the origin of the disease as P*akvasaya* and the cause for the disorder as the *Beejaavayavadusthi*, the main line treatment that can be adopted is the *Vatahara* line of treatment after the removal of *Ama* condition. This treatment protocol was followed in the hospital which showed improvement inmotor development of the child.

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