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Case Study

EFFECT OF AYURVEDIC TREATMENT MODALITIES IN SAHAJA SARVANGAVATAM (INBORN ERROR OF METABOLISM)

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Abstract

A male child of 6 years old attended the Bala roga outpatient department with the following symptoms like difficulty in walking, fine motor skills like pincer grasp not developed, history of developmental delay. He was diagnosed as a case of inborn error of metabolism with pyruvate carboxylase deficiency in modern science. Inborn error of metabolism is an inherited metabolic disease caused due to defect in gene that code for enzyme which facilitates conversion of certain substances into other products. Defect in this gene leads to improper / poor conversion of substances which inturn gets accumulated in various organs affecting its normal functions. The condition was correlated to sahaja vatavyadhi / sahaja sarvangavata in Ayurveda. Good improvements were obtained by Ayurvedic treatment modalities in gross motor and fine motor milestones, speech and cognition in three courses of treatment.

Keywords: Inborn error of metabolism; Sahaja vatavyadhi; Sahaja sarvangavata; Ayurveda.

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INTRODUCTION

Inborn Error of Metabolism (IEM) also known as congenital metabolic diseases or inherited metabolic diseases that arise due to defects in genes that code for enzymes that facilitate conversion of various substances into other products. Mostly these problems arise due to accumulation of substances which are toxic or interfere with normal function or reduced ability to synthesize essential compounds. The inherited metabolic diseases are categorized as disorders of carbohydrate metabolism or lysosomal storage diseases. Inborn error of metabolism lead to enormous number of diseases and wide range of systems are affected.

The manifestations are growth failure, failure to thrive, weight loss, developmental delay, seizures, congenital malformations, hypothyroidism, unusual facial features etc.

In Ayurveda Inborn Error of Metabolism can be considered as sahajavata vyadhi. The normal functions of vata like moulding the shape of embryo, bringing together different parts of the body in garbhavastha (Intrauterine life), prompting all types of actions in body, controlling and impelling mental activities, coordinating all the sense faculties etcare deranged during the growth of foetus itself due to beeja dushti (genetic cause).[1] Deranged vata in garbha (embryo) affects the growth of baby in womb leading to congenital malformations and anomalies of various systems. The diseases caused due to defect in beeja (male and female gamete) are referred to be sahaja vyadhi (genetic disorders) in Ayurveda.[2] Since this disease is due to a defect in gene and the karma of vata is predominantly affected it is diagnosed/named as sahaja vatavyadhi / sahaja sarvangavatam.

CASE HISTORY

A male child of 6 years old attended the Bala roga outpatient department of Govt. Ayurveda Medical College & Hospital, Nagercoil with a O.P. No.: 11687, to undergo 3rd course of treatment, I.P. No.: 301 for the following complaints.

Chief complaints

Difficulty in walking, fine motor skills like pincer grasp not developed, history of developmental delay.

History of present illness

Child was having delayed milestones and poor speech. He is a diagnosed case of inborn error of metabolism with pyruvate carboxylase deficiency. Parents were advised not to take further treatment since there is no hope of improvement in Allopathy for this condition. He came to our hospital at his age of 3 years and 8 months with the following complaints - Inability to sit and stand without support, poorly developed fine motor skills, marked hypotonia, genu recurvatum, difficulty in speech, mild talipes equino varus, umbilical hernia and coarse facies. Child showed good improvement and undergone three courses of treatment.

Birth history

FT, LSCS. B.WT – 2.5kg, delayed cry, poor feeding. H/o. seizure on 10th postnatal day. H/o. RDS. Child is diagnosed to have deficiency of pyruvate carboxylase. Serum lactate – 38.6 mg/dl, S. pyruvate – 1.3 mg/dl and S. ammonia – 83 micro gm/dl.

Gross motor milestones

HC – 1 ½ years
Crawling – 2 years
Sitting with support – 1 ½ years
Sitting without support – 2 years
Standing with support – 3 years
Standing without support – 5 years
Walking with support – 4 years
Walking without support – 5 years

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Personal history

Bowels – once a day; Micturition - 5D/2N; Sleep – sound sleep; Appetite – good; Diet – mixed.

General examination

Poorly nourished, responds well, pallor seen.
No evidence of icterus, clubbing, cyanosis, oedema and lymphadenopathy.

Head to foot examination

Widened space between eyebrows, Protruded eyes, Macroglossia, coarse facies, alopecia, umbilical hernia.

Systemic examination

CNS – Oriented, memory is intact, no disturbances in speech
HMF – Intact
DTR – Grade 2
Cranial nerves and sensory perceptions are intact
MSS – Muscle bulk – reduced in R/L upper & lower limbs, Muscle tone – hypotonic, Muscle power – Grade 4
RS – NBVSH
CVS – No added sounds and S1 & S2 heard
GIT – Umbilical hernia, Slight abdominal protrusion, No organomegaly.
Modern diagnosis – Inborn error of Metabolism (pyruvate carboxylase deficiency)
Ayurvedic diagnosis – Sahaja sarvanga vatam.

Treatment given

Considering the genetic nature of disease, a complete cure is not possible. Following deepana (appetizer) and pachana (digestive) along with vata samana (alleviates vata dosha) treatment repeatedly can arrest progression of disease and can improve the growth and development of the child.

The treatment protocol followed was deepana, paachana, udwartana (powder massage), vicharana snehapaana (oleation) followed by mridu virechana (mild laxative), abhyanga (oil application), siropichu (oil douch on head), kayaseka (pouring of decoction on whole body), shastikapinda sweda (fomentation with a variety of rice), yogavasthi (enema) and nasyam (administering medicine through nostrils).

1. Deepana and paachana – Rajanyadhi churnam – 5 g, BD and Vaishwanara churnam 10 g BD
2. Udwartana with Kolakulathadi churnam for 3 days.
3. Vicharana snehapaanam with Kalyanaka ghritam 10, 20, 30 for three days followed by mridu virechanam (mild laxative) with Trivrt ksheeram – 10 g (Trvrt churnam processed with 50 ml milk). 3 to 5 vegas (bouts) observed.
4. Abhyangam with Dhanwanthara thailam for 7 days
5. Shiropichu with Dhanwanthara thailam for 28 days
6. Kaya sekam with Dhanwanthara thailam for 7 days
7. Shastikapinda swedam for 7 days
8. Yogavasthi with Dwipanchamooladhi vasthi yogam
9. Prathimarsha Nasyam with ksheerabala avarthy.

Internal medication

1. Rajanyadhi churnam – 5 g with ghrtam twice daily
2. Mahakalyanaka ghrtam – 5 g twice daily
3. Dhanwanthara thailam – external application for head and body
4. Saraswatharishtam – 15 ml three times a day.

Follow up – once in two months

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Image 1: Before treatment

Image 2: After treatment

Table 1: Improvements observed after 3 courses of treatment

First course	Second course	Third course
He could able to stand with support; speech improvement seen	Stability in LL improved Able to walk without support Overall gross motor skills improved	Fine motor skills improved

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Table 2: Investigations

Content \ Date	12/01/2010 During neonatal period	30/07/2014 After second course of treatment	16/06/2015 After third course of treatment	Normal reference range
Blood	38.6 mg/dL	23.3 mg/dL	17.2 mg/dL	4.5 – 20 mg/dL
Lactate		2.6 mmol/L	1.9 mmol / L	0.5 – 2.2 mmol / L
Blood NH3	83 microgm / dl		32 micromol /L	17-80 microgm/ dL 11-35 micromol/L

RESULTS

Good improvement in gross motor and fine motor milestones, speech and cognition are observed.

DISCUSSION

It is a case of inborn error of metabolism. The child is found to have pyruvic carboxylase deficiency. It is an inherited disorder that causes lactic acid and other potentially toxic substance to accumulate in the blood. High levels of these substances can damage the body's organs and tissues, particularly in the nervous system. This enzyme is active in mitochondria, involved in several important cellular functions including the generation of glucose – body's main energy source. Pyruvate carboxylase also plays a role in the formation of myelin and neurotransmitters.

In Ayurveda IEM can be considered as sahaja vyadhi. Beejadushti (Abnormality in male/female gamete) causes dhatwagnimandhyam (poor metabolic activity at the level of tissues) which affects dhatu parinama (transformation of tissues) leading to poor status of dhatus (tissues elements) and excessive formation and accumulation of malas (waste products) in the body, associated with tridosadushti (vitiation of three doshas) predominantly vata dosha. This pathology starts in garbha (embryo) itself leading to congenital malformations and anomalies of various systems.

Considering the genetic nature of the disease, a complete cure is not possible. The disease progress and complications can be arrested by following agnideepana, sodhana (purificatory measures) and vata samana treatment procedures repeatedly. Therefore deepana pachana drug was administered initially to augment the digestive capacity and udwartana was done externally for srothosodhanam (clearing the channels), followed by snehapaana and virechana to eliminate the accumulated malas. Since the patient is a child vicharana snehapaana and mridu virechana has been advocated.[4] After purifying the body with mild purificatory measure vata samana procedures like abhyanga,[5] kayaseka and shastikapindasweda has been administered which improved the blood circulation to all parts of body, improved muscle tone, provided nourishment and strengthened the muscles and nerves. Vasthi,[6] the principle treatment for vata administered to eliminate the accumulated doshas / malas in the body and to enhance the functions of Central Nervous System. Shiropichu[7] and nasyam[8] administered to improve the mental activities and to enhance functions of central nervous system. Further administration of oral medicines showed good improvement in signs and symptoms like gross and fine motor milestones and speech.

Mild shodhana karma (purification therapy) was done continually to eliminate the accumulating malas along with oral medication.

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The child has undergone three courses of treatment and the improvements are summarised in Table 1 and changes in the laboratory findings are summarised in Table 2.

CONCLUSION

Inborn error of metabolism can be compared to sahaja sarvanga vatam. Due to genetic nature, the condition cannot be cured. Hence efforts are taken to improve the condition of the child. Here there is sahaja dhatwagnimandhyam causing poor formation of dhatus and excessive accumulation of malas finally affecting vata karma. Therefore proper shodhana and vatashamana treatment along with agni deepana showed good improvement in attaining gross and fine motor milestones and speech.

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