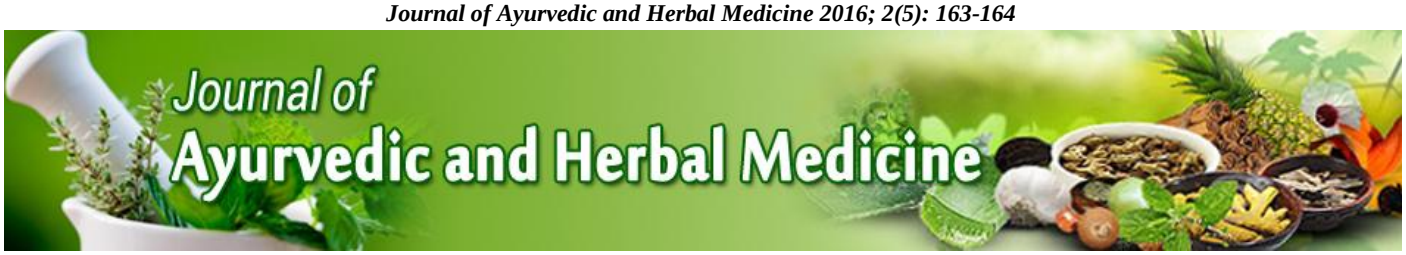

Case Report

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Ayurvedic approach to floating harbor syndrome: A case report

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ABSTRACT

Floating-Harbor syndrome (FHS) is an extremely rare genetic disorder characterized by a distinctive facial appearance, various skeletal malformations, delayed bone age, and expressive and receptive language delays. Children may be below average height for their age (short stature). Floating-Harbor syndrome is a rare disorder; only about 50 cases have been reported in the medical literature. The diagnosis is established by presence of a heterozygous SRCAP pathogenic variant in those with clinical findings of FHS. Early intervention programs, special education, and vocational training to address developmental disabilities; communication rehabilitation with sign languages or alternative means of communication; and behavior management by a behavioral specialist/psychologist with consideration of medication as needed. The present case report is of a two year ten month old male baby who attended the out-patient department of Kaumarabhritya of Shri Dharmasthala Manjunatheshwara College of Ayurveda and Hospital, Udupi, Karnataka, India.

Keywords: Floating-Harbor syndrome, Genetic disorder, Developmental disabilities.

INTRODUCTION

Floating-Harbor syndrome also known as the Pelletier-Leisti syndrome is a very rare genetic disorder characterized by a triad of short stature, speech and psychomotor development delay, and facial anomalies [1]. It was discovered in 1973 by Pelletier and Feingold in a patient admitted at the Boston Floating Hospital, named after the first two identified patients from Boston Floating Hospital and Harbor General Hospital respectively [2]. It is a very rare disorder and to date only twenty confirmed cases have been recorded. Floating Harbor Syndrome is listed as a 'rare disease' by the Office of Rare Diseases (ORD) of the National Institutes of Health (NIH) [3]. This means that Floating Harbor Syndrome, or a subtype of Floating Harbor Syndrome, affects less than 200,000 people in the US population [4]. The mechanism of disease in FHS is suspected to be dominant-negative due to the non-random clustering of truncating mutations in the final exon that result in the loss of the major trans activation function of SRCAP located in a 655 residue C-terminal fragment, evidence that expression of a construct solely consisting of the CBP interaction domain of SRCAP strongly inhibits CREB-mediated trans activation in a dominant-negative fashion, and the existence of patients with haplo insufficiency of SRCAP who do not have features of FHS.

CLINICAL CHARACTERISTICS

Floating-Harbor syndrome (FHS) [5] is characterized by typical craniofacial features; low birth weight, normal head circumference, and short stature; bone age delay that normalizes between ages 6 and 12 years; skeletal anomalies (brachydactyly, clubbing, clinodactyly, short thumbs, prominent joints, clavicular abnormalities); severe receptive and expressive language impairment; hyper nasality and high-pitched voice; and intellectual disability that is typically mild to moderate. Difficulties with temperament and behavior that are present in many children tend to improve in adulthood. Other features can include: hyperopia and/or strabismus; conductive hearing loss; seizures; gastroesophageal reflux; renal anomalies (e.g., hydronephrosis/renal pelviectasis, cysts, and/or agenesis) and genital anomalies (e.g., hypospadias and/or undescended testes).

CASE REPORT

A two year ten month male baby, resident of Mysore district, has attended out-patient department of Shri Dharmasthala Manjunatheshwara College of Ayurveda and Hospital, Udupi, Karnataka, India. The subject was the product of 1st pregnancy to non-consanguineous phenol typically normal parents. At birth, the parental age was 28 and 33 years for mother and father respectively. On attending the out-patient department, the patient presented with delayed miles stones since birth (mainly with delay in walk and

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speech). The patient was said to be born through LSCS (failed induction and meconium stained). The baby cried immediately after birth but the cry was said to be feeble and there was history of NICU admission. Examination at birth revealed weight 2.8 kg, length 47 cm and occipito-frontal circumference 30 cm. The developmental status/history is detailed in table 1. The clinical features presented by the patient are detailed in table 2. The anthropometry measurement on the day of admission is detailed in table 3. There was a normal karyo typing report and FISH study report. Looking the main features baby got diagnosed as floating harbour syndrome.

Table 1: Developmental status/history

Milestone	Attained age	Normal limit
Neck control	6month	3rd month
Pull to sit up	1 year	4th month
Sit without support	1 yr 6 month	7-8th month
Stand holding	1 yr 10 month	9-10th month
Social Smile	1 ½ month	3rd month
Becomes alert when mother is around	2nd month	2-3 months
Reaching for dropped object	7th month	7-8th month
Laughs for a peek a boo game	8th month	8-9 months
Holds rattle	8th month	4-5 months
Holds object in each hand	10th month	7th month
Babble	7th month	5-6 months
Monosyllable	9th month	8-9 months
Bisyllables	2year and 6month	10-11 months

Table 2: Clinical features presented by the patient

S. No.	Areas	Clinical features
1.	Head	Small forehead
2.	Hands (Dermatoglyphics)	Simian crease
		Brachydactyly
		Distal 't' triradius`
		Increased axial tri radius angle
3.	Face	Dysmorphic face
		Triangular face
		Prominent beaked nose with broad columella
		Prominent eyes
		Hypertelorism
		Micrognathia
		Folded cupped ears
4.	Heart	Patent ductus arteriosus
		Ventricular septal defect
5.	Genitalia	Hypospadias and bifid scrotum
6.	Abdomen	Cafe-au-lait spots one on abdomen
7.	CNS	Mental retardation
		Developmental delay
8.	Musculoskeletal System	Hypertonia
		Cafe-au-lait spots on right mid arm

Table 3: Anthropometry measurement on the day of admission

Anthropometry	Measurement in patient	Normal measurement
Head circumference	46cms	49cms
Chest circumference	48cms	52cms
Height	82cms	95cms
Weight	9 kg	14kg
Mid arm(Right)	15cms	16cms
Mid arm(Left)	15cms	16cms

OBSERVATIONS AND DISCUSSION

As per Ayurvedic parlance, the condition can be better understood as, Sahaja beeja-beeja bhaga-beeja bhaga avayava janya vikara [6]. For the same mainly vatahara treatments is to be resorted to. Hence here also Vatahara kriyakramas were carried out such as Sarvanga abhyanaga with Ashwagandha bala laskshadi taila, Shastika shali pinda sweda and Matra basti with Samvardhana ghrita [7] .The shamana aushadis such as Syrup Intellect, Medya vati and Kumara kalyana Rasa were administered. Added to these, physiotherapy was also advised. The treatment was carried out for a period of seven days. Later the patient was advised follow up of 14 days during which the patient was adviced with medicines such as Syrup Intellect (5ml bd), Medya vati (1 bd) and Kumara kalyana Rasa[8] (1/4th tab bd) along with Sarvanga abhyanga with Ashwagandha bala laskshadi taila. The improvement achieved by the patient during the treatment and follow up period is detailed in table 4.

Table 4: Improvements achieved by the patient throughout the treatment and follow up period

S. No.	Visiting date	Improvement recorded
1.	11/05/2016 to 27/05/2016	1. Slight increase in strength of all four limbs
2.	01/06/16	1. Started standing with support 2. Can stand without support for two minutes 3. Activity and response increased.

CONCLUSION

Floating Harbor syndrome is a very rare dysmorphic/mental retardation syndrome affecting both gender but more frequent among the female gender. Most of the reported cases occur sporadically, but a few familial cases have been reported, raising the possibility of autosomal dominant mode of inheritance. The treatment protocol adopted in this case showcases a very effective approach in improving the quality of life of the child. Hence the same treatment plan can be adopted for similar diagnosed cases.

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