



Case Study

## AYURVEDIC APPROACH TOWARDS WOLF-HIRSCHHORN SYNDROME: A CASE REPORT

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### ABSTRACT

Wolf-Hirschhorn Syndrome is a rare chromosomal disorder caused by the partial deletion (structural aberration) of the short arm of chromosome 4. It is also known as 4P syndrome. The disease was first reported by Cooper, Hirschhorn and Wolf. The characteristic features of the patient depend on the extent of deletion of the genetic materials. The patient has craniofacial dysmorphism and the dysmorphic features are collectively known as Greek Warrior Helmet Facies. There is no direct correlation available for Wolf-Hirschhorn Syndrome in Ayurveda. Since it is a congenital pathology occurring due to chromosomal aberration it may be understood as a *Sahaja vyadhi* caused due to *Beeja dosha*. Here in this case the micro/partial deletion of genetic materials from short arm of chromosome 4 is due to the *Beeja dosha* that has occurred during the formation of *Garbha* (zygote). This *Beeja dosha* is responsible for the manifestation of different presentation in Wolf-Hirschhorn Syndrome which can be understood as a *Vyadhi Sankara* i.e., syndrome. The patient was presented with the features of facial dysmorphism, hypospadias, developmental delay and seizures. Hence the case was diagnosed as *Vyadhija Phakka* with *Vataja Apasmara*. In the present case the focus was mainly in the correction of developmental delay and to prevent the relapse of seizures. An integrated treatment protocol comprising *Phakka chikitsa* and *Vataja Apasmara chikitsa* were administered. Though there was no change in the facial dysmorphisms, but significant improvements were observed in the gross motor, language and communication milestones after the treatment.

### INTRODUCTION

Wolf-Hirschhorn Syndrome is a rare chromosomal disorder caused by the partial deletion (structural aberration) of the short arm of chromosome 4. It is also known as 4P syndrome. The disease was first reported by Cooper, Hirschhorn and Wolf.<sup>[1]</sup> The characteristic features of the patient depends on the extent of deletion of the genetic materials. The patient has craniofacial dysmorphism and the dysmorphic features are collectively known as Greek Warrior Helmet Facies. The common phenotype abnormalities include microcephaly, frontal bossing, high frontal hairline, prominent glabella,

hypertelorism, ptosis, iris coloboma, wide nasal bridge, down slanting palpebral fissures, strabismus, exotropia, carplike mouth, large misshapen ears, skeleton abnormalities, hypospadias and kryptorchismus. Dysmorphic features are also associated with hypotonia, seizures and reliable insensitiveness to pain. There is no definite cure for Wolf-Hirschhorn Syndrome. Life expectancy has not been well documented but one third of patients die in the first year due to pneumonia or congestive heart failure. Only few patients survive into the first decade.<sup>[2]</sup> In Ayurveda it is understood as a *Sahaja Vyadhi* caused due to *Beeja dosha* during the formation of *Garbha* resulting in a *Vyadhi Sankara*. *Vyadhija Phakka* and *Vataja Apasmara* were mainly included in the *Vyadhi Sankara* presentation. *Vyadhija Phakka lakshanas* like *Pramlaana adhara Kaya*, *Ksheena mamsa bala*, *Samshushka shiro mukha* were observed in this patient.<sup>[3]</sup> *Vyadhija Phakka* and *Vataja Apasmara chikitsa* were administered to the patient.

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**Case Report**

A 2 year 9 month old baby was brought to the Bala Roga (Pediatric) OPD of Sri Jayendra Saraswathi Ayurveda College and Hospital, Chennai with complaints of developmental delay (all the 4 domains) and facial dysmorphism. On examination of the child there was microcephaly, wide nasal bridge, frontal bossing, large misshapen ears and hypertelorism. The child was the first born to young and healthy parents having second degree consanguinity. The family history was positive. The baby was born at 43<sup>rd</sup> week under normal vaginal delivery with a birth weight of 2.57kg. The baby cried approximately 3 minutes after birth as per the words of the mother. There were no antenatal complications for the mother. There was no

h/o neonatal jaundice. After 25 days of birth the baby was admitted in NICU for 15 days due to UTI as per the mother and was diagnosed with hypospadias. There was a h/o partially treated pyogenic meningitis for the baby at the 7<sup>th</sup> month. The baby was exclusively breast fed till the age of 6<sup>th</sup> month and continued breast feeding till 1 & half years. The status of the child with respect to the milestones at the time of admission were the child was not able to stand without support, not able to walk alone, unable to wave bye-bye, and language/communication milestone was achieved only till cooing. There was previous history of seizures and the last episode of seizure was reported on August 2022. The child is under regular antiepileptic medication - Levipil -2ml - 0-2ml A/F

History of Milestones Achievement		
Sr. No	Miles stones	Attained Age
1	Neck holding	1 year
2	Rolls over	5 months
3	Sits without support	1 year 4 months
4	Stands with support	2 years
5	Stands without support	Not yet achieved
6	Walks alone	Not yet achieved
7	Bidextrous grasp	8 months
8	Unidextrous grasp	12 months
9	Social smile	1 year
10	Recognizes mother	1 year
11	Stranger anxiety	2 years
12	Wave bye-bye	Not yet achieved completely
13	Comes when called	2 years
14	Alert to sound	1 year
15	Cooing	1.5 years
16	Mono syllables	1.5 years
17	Bisyllables	Not yet achieved

**Investigations**

Karyotype analysis - 46 XY

MRI Brain (done on 2022) - Corpus callosum shows mild thinning with undulating margins

MR Venogram (done on 2022) - Hypoplastic left transverse sinus

**Diagnosis**

Modern Diagnosis - Wolf-Hirschhorn Syndrome (clinical diagnosis, suspected on karyotype analysis)

Ayurveda Diagnosis - *Sahaja Vyadhi + Vyadhija Phakka*

**Management**

Sr. No	Date	Treatment
1	27.03.23 -30.03.23	<ul style="list-style-type: none"> <li><i>Ruksha churna Pinda swedam with Masha churna +Kulatha Churna</i></li> <li><i>Shiro talam with Rasna churnam + Ashwagandha Churnam + Brahmi Churnam + Amalaki Churnam +Brahmi Tailam</i></li> </ul>

		<ul style="list-style-type: none"> <li>• <i>Matra Basthi with Kalyanaka ghritam 20ml + 2-3 pinch of Vacha Churnam</i></li> </ul>
2	31.03.23 -06.03.23	<ul style="list-style-type: none"> <li>• <i>Sarvanga Abhyanga with Dhanwantaram tailam</i></li> <li>• <i>Shashtika Shali Pinda Swedam</i></li> <li>• <i>Shiro Pichu - Dhanwantaram +Brahmi tailam for 45 minutes</i></li> <li>• <i>Matra Basthi with Kalyanaka ghritam 20ml + 2-3 pinch of Vacha Churnam</i></li> <li>• <i>Pratimarsha Nasyam with Kalyanaka Ghritam – 1 drop (in the evening)</i></li> </ul>

### Oral Medications

- *Vacha churnam + Rudraksha Churnam +Rajanyadi Churnam + Kalyana Avaleha Churnam*– ½ tsp twice daily before food with honey
- *Dhanwantaram Gutika* – 1-1-1/ B/F
- *Smruti Sagara Rasa* – ½ -0-1/2 A/F
- *Syrup Cognium* – 5ml - 0 - 5ml A/F

### Outcome

Sr. No	Milestones (BT)	Milestones (AT)
1	Not able to stand without support	Stands without support
2	Walks only with support	Walks without support
3	Unable to climb up and down stairs	Climb up and down stairs by holding the rail with one foot per step.
4	Unable to waves bye-bye	Partially trying to wave bye-bye
5	Speech – only cooing	Speech - laughs loudly and tries to speak monosyllables

### Discharge Medications: (For 1 month)

1. *Kalyanaka Ghritam (1tsp) + Vacha churnam + Rudraksha Churnam +Rajanyadi Churnam - Kalyana Avaleha Churnam ( ½ tsp) twice daily before food*
2. *Dhanwantaram Gutika* – 1-1-1/ B/F
3. *Smruti Sagara Rasa* – ½ - 0 -1/2 A/F
4. *Syrup Cognium* – 5 ml - 0 - 5 ml A/F
- *Shiro talam - Rasna churnam +Ashwagandha Churnam + Brahmi Churnam + Amalaki Churnam + Brahmi Tailam (in the evening)*
5. *Sarvanga Abhyanga with Dhanwantaram tailam (1/2 hr before bath) followed by warm water bath.*

### DISCUSSION

Wolf-Hirschhorn Syndrome leading to developmental delay can be understood in Ayurveda as *Vyadhija Phakka Roga* caused due to a *Sahaja Vyadhi*. Wolf-Hirschhorn syndrome can be brought under the heading of *Sahaja vyadhi* as there is chromosomal aberration due to micro deletion from the short arm of chromosome 4. Here the cause is *Beeja dosha* for *Sahaja Vyadhi* (Wolf-Hirschhorn Syndrome) and *Jwaradi* (Pyogenic Meningitis) mentioned as a *Nidana* for *Vyadhija Phakka* were also present in this case. The *Lakshanas* of *Vyadhija Phakka* like *Pramlaana adhara Kaya*, *Ksheena mamsa bala*,

*Samshushka shiro mukha* were observed in this patient.<sup>[4]</sup> Seizures are one of the prominent clinical feature of this syndrome. Hence the treatment was focused mainly in correcting the developmental delay as well as to prevent the recurrence of seizures. Here the seizures may be understood as *Vataja Apasmara*. Here in this case *Apasmara* is one of the presentations of *Sahaja Vyadhi*. So an integrated treatment protocol for *Vaydhija Phakka* and *Vataja Apasmara* was administered for a period of 10 days. The MRI finding – mild thinning of Corpus Callosum can be understood as reduction in *Sthoolata* and *Sthira guna* which is an attribute of *Kapha* due to increased *Ruksha guna* of *Prana Vata*. Hence *Brumhana chikitsa* was planned for the *Shiras* and *Kaya* which helped the child in achieving the milestones. The history of seizures was kept in mind while selecting the medicines. Initially a *Mrudu rukshana* was advised in the form of *Ruksha churna pinda sweda* with *Masha* and *Kulatha churnam*. *Shiro talam* was started with *Brahmi tailam* and a combination of *Rasna churnam*, *Ashwagandha Churnam*, *Brahmi Churnam* and *Amalaki Churnam*. *Brahmi tailam* was judiciously selected as it's a *Medhya dravya* and *Brahmi tailam* is also proved to be effective for *Apasmara*. To prevent the *Vata prakopa*, *Churnas* of *Rasna ashwagandha* were selected along with *Brahmi tailam*. *Matra Basthi* was started from the day 1 with

*Kalyanaka Ghritam* and *Vacha Churnam*. *Kalyanaka Ghritam* was selected due to the predominance of *Vata dosha (Prana Udana and Vyana)* in this case and also it is beneficial in improving the speech by correcting the function of *Prana Vata* and *Udana Vata*. *Kalyanaka ghritam* is also mentioned in the *Chikitsa* of *Vyadhija Phakka* for *Snehanam*. Moreover *Basthi* is helpful in *Vataja Apasmara Chikitsa* thereby helped in the prevention of relapse of seizures.<sup>[5]</sup> After 4 days of *Rukshana*, the *Chikitsa* was shifted to *Brumhana* for *Shiras* and *Kaaya*. *Sarvanga Abhyanga* was done with *Dhanwantaram tailam* for *Sheeta brumhanam* followed by *Shashtika Shali Pinda Swedam*. It helped in strengthening the skeletal muscles and improving the tone thereby helped in achieving the delayed motor milestones. *Murdhni tailam* was administered in the form of *Shiro Pichu* with *Dhanwantaram* and *Brahmi Tailam*. *Partimarsha Nasyam* was administered with 1 drops of *Kalyanaka Ghritam*. *Nasyam* and *Murdhni tailam* was mainly administered for the correction of thinning of corpus callosum. Internally *Smruti Sagara Rasa* mentioned in the context of *Apasamara chikitsa* was administered to prevent the relapse of seizures and it also acts as *Medhya rasyana* due to its *Bhavana dravyas* i.e., *Vacha swarasa* and *Brahmi swarasa*. Cognium syrup containing *Brahmi*, *Shankapushpi* etc was also administered as a *Medhya rasayana* for a period of 1 month after discharge.

## CONCLUSION

Wolf-Hirschhorn Syndrome in Ayurveda can be understood as a *Sahaja Vyadhi* caused due to *Beeja dosha* resulting in *Vyadhija Phakka* with developmental delay as the major feature. In the

present case the focus was mainly in the correction of developmental delay and to prevent the relapse of seizures. An integrated treatment protocol comprising *Phakka chikitsa* and *Vataja Apasmara chikitsa* were administered. Significant improvements were observed in the milestones after the treatment. Hence in chromosomal conditions like Wolf-Hirschhorn Syndrome, though there is a limitation in correcting the cause and the facial dysmorphisms, developmental delay and relapse of seizures can be safely and effectively managed with a well framed Ayurvedic treatment protocol.

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