Publisher
Maharshi Charaka Ayurveda
www.maharshicharaka.in

E-ISSN:2456-3110

Journal of Ayurveda and Integrated Medical Sciences

2025 Volume 10 Number 5 MAY

Case Report

Angelman Syndrome

Check for updates

Exploring Integrative Ayurveda in the Supportive Management of a Child with Angelman Syndrome - A Case Study

Meghana SK[1*], Girish Kumar SV[2], Meghana V[3]

DOI:10.21760/jaims.10.5.44

[1*] Meghana SK, Final Year Post Graduate Scholar, Department of Kaumarabhritya, Shri Dharmasthala Manjunatheshwara Institute of Ayurveda and Hospital, Bengaluru, Karnataka, India.

[2] Girish Kumar SV, Associate Professor, Department of Kaumarabhritya, Shri Dharmasthala Manjunatheshwara Institute of Ayurveda and Hospital, Bengaluru, Karnataka, India.

[3] Meghana V, Final Year Post Graduate Scholar, Department of Kaumarabhritya, Shri Dharmasthala Manjunatheshwara Institute of Ayurveda and Hospital, Bengaluru, Karnataka, India.

Angelman Syndrome (AS) is a rare neurogenetic disorder caused by the pathological lack of expression of the UBE3A gene on the maternal chromosome, combined with the physiological genomic imprinting or silencing of the paternal chromosome in neurons. It clinically presents with Intellectual disability, hypotonia, inappropriate laughter, poor speech etc,. The estimated prevalence is 1 in 16,000 live births. Though it presents significant challenges to affected children and their families, early intervention and proper care can improve the quality of life. A case of 5 year old female child with k/c/o Angelman Syndrome visited OPD, Department of Kaumarabhritya, Shri Dharmasthala Manjunatheshwara Institute of Ayurveda and Hospital, Bengaluru. She was presented with slurred speech with minimal words (Vak Vikara), lack of social communication, hyperactivity and inattentiveness (Chanchala Chitta), reduced concentration (Anavasthita Chittatva), unsteady gait and hypotonia, sleep disturbance. Due to involvement of Genetic susceptibility status and Predominant Vata Dosha symptoms, the condition is diagnosed as Beejadushtigata Vata Nanatmaja Vikara and appropriate Shamanaoushadi and Panchakarma therapies. Significant improvements were seen after the treatment.

Keywords: Angelman syndrome, Beejabhaga Dushti, Beejabhagavayava Dushti, Panchakarma therapies, Vata Nanatmaja Vikara

Corresponding Author

Meghana SK, Final Year Post Graduate Scholar, Department of Kaumarabhritya, Shri Dharmasthala Manjunatheshwara Institute of Ayurveda and Hospital, Bengaluru, Karnataka, India.
Email: meghanask78@gmail.com

How to Cite this Article

Meghana SK, Girish Kumar SV, Meghana V, Exploring Integrative Ayurveda in the Supportive Management of a Child with Angelman Syndrome - A Case Study. J Ayu Int Med Sci. 2025;10(5):281-287.
Available From
https://jaims.in/jaims/article/view/4319/

To Browse

Manuscript Received	Review Round 1	Review Round 2	Review Round 3	Accepted
2025-04-09	2025-04-24	2025-05-06	2025-05-14	2025-05-24

Conflict of Interest	Funding	Ethical Approval	Plagiarism X-checker	Note
None	Nil	Not required	12.46	

Introduction

Angelman Syndrome is a neurodevelopmental disorder caused by maternal micro-deletion of chromosome 15q11-13 or paternal uniparental disomy, leading to mutation or dysfunction of UBE3A gene. This gene plays a critical role in cellular protein management and nervous system development. Hence the prominent features involving learning and memory impairement. In further it is clinically characterized by Severe intellectual disability, hypotonia, fair hair and skin, midface hypoplasia, ataxia, tremulous movements, large mouth, frequent drooling, inappropriate laughter, feeding difficulties, gastroesophageal reflux, poor or absence of speech, sleep disturbances. The estimated prevalence is 1:16,000 live births.**[1]** While there is no definitive cure for Angelman syndrome, but physiotherapy, behavioural therapy, occupational, speech therapy and anti-epileptic medications are recommended for symptomatic management. In *Ayurveda*, considering its genetic susceptibility, Angelman syndrome can be correlated with *Beeja Dushtigata Vikara*. More specifically, it aligns with *Beeja Dushtigata Vata Prakopa Janya Vikara,* due to the predominance of *Vata Dosha,* along with the involvement of *Pitta* and *Kapha Dosha*. The *Ayurveda* manifestations include *Vak Vikruti* (speech disorder), *Mamsa Asthirata* (hypotonia, writing issues)*, Gati Vaishamya* (Unsteady gait), *Chanchala Chitta* (inattentiveness and hyperactivity), *Nidra Vikruti* (irregular sleep pattern), *Medha Vikara* (Intellectual disability). Ultimately it can be considered as *Beejadushtigata Vatananatmaja Vikara.***[2]**

Though the *Beejadushti* is considered as *Asadhya*, symptomatic management is possible through *Panchakarma* therapies and *Shamanaoushadi*. Emphasis is placed on *Vata Shamana Chikitsa* due to the predominance of *Vata*, and due to the involvement of *Beejadushti*, *Swarna* preparations may be beneficial.

Case Report

Pradhana Vedana

A 5 year old female child was brought to our OPD by her parents. The informants c/o slurred speech with minimal words, non-verbal communication, hypotonia, unsteady gait, writing issues, Feeding difficulty, frequent inappropriate laughter, irregular sleeping pattern, unable to maintain peer relationship, lack of reciprocation, hyperactivity, inattentiveness (in commuinty and home) and reduced concentration in studies since 2 years.

H/o Seizures at 6 month of life.

Vedana Vruttanta

The first preterm child (born at 36 weeks) to non-consanguineous parents through IVF was delivered through LSCS, cried immediately after birth and weighing 2 kg. Child was said to be normal and attained milestones as per the chronological age, till the age of 6 months of age. After which she developed a high grade fever accompanied by seizures.

The child was on medication from 6 months to 11 months of age, during which the parents observed a lack of neck control alon with difficulties in both bidextrous and unidextrous grasp. After 11 months, the child gradually achieved better neck stability and improved dextrous grasp. In later, although she developed the ability for pincer grasp, child predominantly continued using a dextrous grasp.

As the child grew older, particularly from around 3 years of age, her parents started noticing behavioural issues, including inattention, poor peer relation, slurred speech and unsteady gait. For these concerns, the parents consulted to Higher centre, where genetic analysis was advised along with therapies.

The child was enrolled in Occupational therapy, behavioural therapy and speech therapy. Based on the genetic analysis report, condition was diagnosed as Angelman syndrome. However, as the parents did not notice significant improvements, they sought further evaluation and management at SDM hospital, Department of Kaumarabhritya, Bengaluru.

Clinical Findings

Developmental

- Slurred speech with minimal words usage and no clarity in speech
- Presence with pincer grasp but grabs with whole hand to scribble

Muscular

- Hypotonia
- Unsteady gait (ataxic?), needs help with stairs

Behavioural issues

- Hyperactivity and Inattentiveness - pays attention for 2 mins, impulsive with behaviour in community and home
- Short attention span
- Stubborn - when she gets avoided for being overly persistent
- Inappropriate frequent laughter
- Sleep disturbances - disrupted sleep pattern causes irritability (on intake of sweet – drowsy)

Communication

- Lack of peer relationship, lack of reciprocation, can follow two step commands, requires multiple repetition for sentence commands.

Clinical Diagnosis

K/c/o Angelman syndrome

Chromosome Analysis by FISH method (03/04/21) : Microdeletion of the region 15q11-13 not seen in any of the cells studied.

Diagnostic Assessment

Table 1: Assessment done by CGI-S-AS (Clinical Global Impression Severity Scale)[3]

Before treatment

Domain	CGI-S-AS Score	
Behavior	Markedly impaired	Severity 5
Fine motor	Mildly impaired	Severity 3
Gross motor	Mildly impaired	Severity 3
Expressive Communication	Mildly impaired	Severity 3
Receptive Communication	Mildly impaired	Severity 3
Sleep	Borderline impaired	Severity 2

Therapeutic Interventions:

Table 2 : *Panchakarma* Procedures adopted during the 1st sitting of IPD admission (18/11/2023 to 27/11/2023)

1st Sitting Treatment			
18/11/2023 & 19/11/2023	20/11/2023 to 22/11/2023	23/11/2023 & 24/11/2023	25/11/2023 to 27/11/2023
Utsadana	Sarvanga Abhyanga	Sarvanga Abhyanga	Sarvanga Abhyanga
Parisheka	Parisheka	Churna Pinda Sweda	Parisheka
Chitrakadi Vati 1/2-0-1/2 B/F	Shirodhara, Matrabasti, M G Upanaha, Asyapratisarana		

Treatment

- *Utsadana* with *Kolakulathadi Choorna* and *Triphala Choorna* mixed with *Asanabilwadi Taila*
- *Parisheka with Dashamoola Kwatha*
- *Shirodhara* with *Dashamoola Ksheerapaka*
- *Sarvanga Abhyanga with Asanabilwadi Taila* and *Ashwagandha Bala Lakshadi Taila*
- *Masha Godhumadi Upanaha* for B/L Lower limbs
- *Matra Basti* with *Saraswatha ghrita* and *Ashwagandha ghrita* - 15ml each
- *Asyapratisarana* with *Suvacha Churna*, *Saraswatha Ghrita*, honey

Discharge advice:

- *Kumarakalyana Rasa* 1/2-0-1/2 with *Saraswatha Ghrita*5ml-0-2.5ml B/F
- *Saraswatharishta* + *Balarishta* - 5ml each with 10ml water (1-0-1 A/F)
- Continue *Asyapratisarana* (*Suvacha Churna*, *Saraswatha Ghrita*, honey)

For 1 month

Table 3 : *Panchakarma* Procedures adopted during the 2nd sitting of IPD admission (22/12/2023 to 31/12/2023)

2nd Sitting Treatment	
22/12/2023 & 23/12/2023	24/12/2023 to 31/12/2023
Utsadana	Sarvanga Abhyanga
Parisheka	Shashtika Shali Pinda Sweda
Chitrakadi Vati 1/2-0-1/2 B/F	Shirodhara, Katipichu, M G Upanaha, Matrabasti, Asyapratisarana

Treatment

- *Utsadana* with *Kolakulathadi Choorna* and *Triphala Choorna* mixed with *Asanabilwadi Taila*
- *Parisheka with Dashamoola Kwatha*
- *Shirodhara* with *Brahmi Taila*
- *Sarvanga Abhyanga* with *Asanabilwadi Taila* and *Ashwagandha Bala Lakshadi Taila*
- *Masha Godhumadi Upanaha* for B/L Lower limbs
- *Katipichu* with *Ashwagandha Bala Lakshadi Taila*
- *Matra Basti* with *Saraswatha ghrita* and *Ashwagandha Ghrita* - 15ml each
- *Asyapratisarana* with *Suvacha Churna*, *Saraswatha Ghrita*, honey

- Physiotherapy

Discharge advice:

- *Kumarakalyana Rasa* 1/2-0-1/2 with *Saraswatha Ghrita*5ml-0-2.5ml B/F
- *Saraswatharishta* + *Balarishta* - 5ml each with 10ml water (1-0-1 A/F)
- Continue *Asyapratisarana* (*Suvacha Churna*, *Saraswatha Ghrita*, honey)
- Continue *Abhyanga* with *ABL Taila and Asanabilwadi Taila*
- Continue Physiotherapy

For 1 month

Table 4 : *Panchakarma* Procedures adopted during the 3rd sitting of IPD admission (10/02/2024 to 19/02/2024)

3rd Sitting Treatment	
10/02/2024 & 11/02/2024	12/02/2024 to 19/02/2024
Utsadana	Sarvanga Abhyanga
Parisheka	Shashtika Shali Pinda Sweda
Sadhyovirechana with Trivrut Leha (30g) f/b Draksha Rasa (100ml)	Shirodhara, Merubasti, Matrabasti, M G Upanaha, Asyapratisarana

Treatment

- *Utsadana* with *Kolakulathadi Choorna* and *Triphala Choorna* mixed with *Asanabilwadi Taila*
- *Parisheka* with *Dashamoola Kwatha*
- *Shirodhara* with *Brahmi Taila*
- *Sarvanga Abhyanga* with *Asanabilwadi Taila* and *Ashwagandha Bala Lakshadi Taila*
- *Masha Godhumadi Upanaha* for B/L Lower limbs
- *Merubasti* with *Ashwagandha Bala Lakshadi Taila*
- *Matra Basti* with *Saraswatha Ghrita* and *Ashwagandha Ghrita* - 20ml each
- *Asyapratisarana* with *Suvacha Churna*, *Saraswatha Ghrita*, honey
- Physiotherapy

Discharge advice:

- *Kumarakalyana Rasa* 1/2-0-1/2 With *Saraswatha Ghrita*5ml-0-2.5ml B/F
- *Mashabaladi Kashaya* + *Ashtavarga Kashaya* – 5ml Each With 10ml Water (1-0-1 A/F)
- Continue *Asyapratisarana* (*Suvacha Churna*, *Saraswatha Ghrita*, Honey)
- Continue *Abhyanga* with *ABL Taila* and *Asanabilwadi Taila*
- Continue Physiotherapy

For 1 month

Follow-Up and Outcomes:

Table 5 : Improvements observed after each sitting, are as follows:

At the end of 1st sitting treatment	At the end of 2nd sitting treatment	At the end of 3rd sitting treatment
▪ Speech improving (able to initiate words) ▪ Socialization improving (plays with kids)	▪ Communication and peer relationship improved ▪ Writing skills improving (attained pincer grasp) ▪ Academic performance improved	▪ Hyperactivity reduced (sits in one place) ▪ Attention span improved ▪ Speaks 4-5 words, forms sentences ▪ Reciprocates to the questions ▪ Walking improved (Steady walk, doesn't need support even in stairs) ▪ Child is performing in a group dance event

Table 6 : Assessment done by CGI-I-AS (Clinical Global Impression Improvement Scale)[3] After treatment

Domain	CGI-I-AS Score	
Behavior	Concentration improved, child started to socialize, hyperactivity reduced, activities improved	2 = much improved
Fine motor	Writing issues improved	1 = very much improved
Gross motor	stability improved, can walk individually in stairs	1 = very much improved
Expressive Communication	Understands the emotions, comes home and expresses the daily activity of school	1 = very much improved
Receptive Communication	Sentence formation improved, reads well	1 = very much improved
Sleep	Doesn't sleep in the afternoon, but still has sleep impairement-feels drowsy after having sweet	2 = much improved

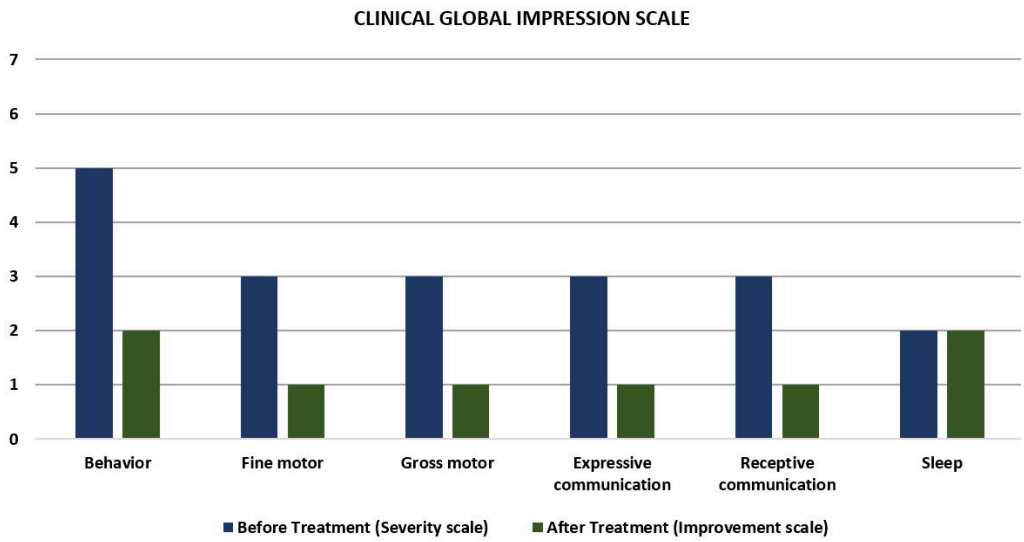

Figure 1: Effect of overall treatment accessed by CGI-Scale

Table 7: To assess of the Quality of life by Barthel Daily Activity Index (after 3 sittings)

Parameters	Before Treatment Score	After 1st Sitting	After 2nd Sitting	After 3rd Sitting
Total Score	20/100	25/100	40/100	60/100

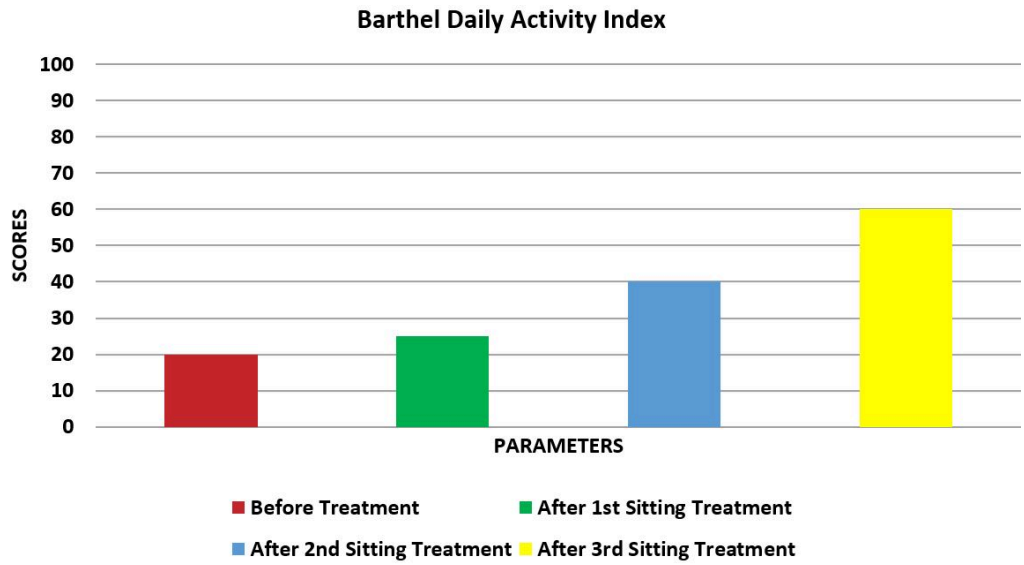

Figure 2 : Assessment of the Quality of life by Barthel Daily Activity Index (after 3 sittings)

Discussion

Angelmen Syndrome can't be exactly correlated with any conditions in *Ayurveda*, but based upon the symptoms, indicates predominance of *Vata* along with *Beejadushti*. Hence present clinical condition case can be correlated with *Beejadushtigata Vata Nanatmaja Vikara*.

The microdeletion of chromosome and defect in UBE3A gene indicates *Beejabhaga Dushti* and *Beejabhagavayava Dushti* which is responsible for cellular protein management and nervous system development.

As per mother history, she had repeated abortions which can be taken as *Garbhashaya Dushti* and at the same time during the *Garbha Avastha*, factors like *Vatakara Ahara Vihara*, *Garbhini Aparipathya Palana* or exposure to *Garbhopaghatakara Bhavas* can also adversely influence the psychological and physical status of the child, which is evident in the present clinical study.

The *Prakupita Vata* (specifically *Prana* and *Udana*) in the *Garbha Avastha,* which further vitiates *Sadhaka* pitta, gets *Sthanasamsraya* in *Shiromarma*, resulting in *Vak Vikruti,* irregular sleeping pattern, inappropriate laughter, hypotonia, hyperactivity, inattentiveness and reduced concentration.

This also involves disturbances in the *Manas*, *Indriya* and *Indriyartha Sannikarsha* leading to impaired *Jnanotpatti* ultimately leading to *Prajnaparadha*, this manifests as poor peer relationship, lack of reciprocation. Hence, the condition reflects the predominant *Vata* impairement, in association with *Prakupita Pitta*, which requires *Vata Shamana Chikitsa*.

***Kumarakalyana Rasa*[4]** is a *Khalviya Rasayana* which contains *Swarna Bhasma* and primarily prescribed in paediatric practice. It *Tridosha Shamaka* and the *Saptadhatu Poshaka* action of *Kumarakalyana Rasa* provides physical and mental strength. ***Swarna Bhasma*[5,6]** has immunodulatory, nootropic action and it is a nervine tonic. It improves overall natural function in the body, improves muscle strength especially acts on nerves, brain, lungs and mind.

Some of the *Panchakarma* therapies adopted during IPD admission are ***Abhyanga*** which helps in pacifying *Vata* and provides nourishment to the body. *Abhyanga* with *Asanabilwadi Taila*[7] and *Ashwagandha Bala Lakshadi Taila,*[8] both of which are *Nadi Balya*, *Shirohitam* and *Vata Shamaka*, helps improve muscle tone and reduces contractions, ultimately enhancing motility and strength. ***Shirodhara*** has been performed using *Dashamoola Ksheerapaka,*[9] where *Dashamoola*, known for its *Vata Shamaka* properties, combined with *Ksheera*, addresses both *Prakupita Vata* and *Pitta,* thus helping to alleviate hyperactivity and sleep disturbances. Additionally, *Brahmi Taila* known for its *Medhya* and antihyperactive effects, has also been used. ***Matra Basti*** with *Saraswatha Ghrita*[10] is known to enhance *Vak, Medha, Smrithi* and *Agni*, which is beneficial in neurocognitive and developmental disorders and *Ashwagandha Ghrita*[11] is indicated in *Vatika Vyadhi*, and known for its *Buddhi* and *Medhya* enhancing property, plays a vital role in managing hyperactivity, inattentiveness, unsteady gait all of which are linked to brain function.

Conclusion

Angelman syndrome is an incurable disease, due to the presence of *Beejadushti*. There is no exact correlation in *Ayurveda* but based on the symptoms, it can be compared with *Vata Nanatmaja Vikara,* and can be managed symptomatically.

By certain *Panchakarma* procedures, *Vata Shamana*, *Medhya* and *Rasayana* line of *Chikitsa* and here *Swarna* preparations plays an important role by there Nootropic effect, *Pushti Prakashi* and *Saukhya* in nature.[5,6]

Acknowledgement: The author extends heartfelt thanks to the child and their family for their cooperation and active involvement in the treatment and documentation process.

References

1. Barkoudah E, Kliegman RM, St Geme JW, Blum NJ, Tasker RC, Wilson KM, et al. Nelson Textbook of Pediatrics. 22nd ed. Canada: Elsevier; 2024. *[Crossref][PubMed][Google Scholar]*

2. Laxmidhar D, editor. Charaka Samhitha of Agnivesha. Sutrasthana, Ch. 20. *1st ed. Varanasi: Chaukamba Krishnadas Academy; 2012 [Crossref] [PubMed][Google Scholar]*

3. Busner J, Targum SD. The Clinical Global Impressions Scale. Psychiatry (Edgmont). 2007 Jul;4(7):28–37. *[Crossref][PubMed][Google Scholar]*

4. Bhala RR. Ayurveda approaches for Bala Roga with special reference to importance of classical formulations. Himalayan J Health Sci [Internet]. 2022 Jun 15 [cited 2022 Jun 15];7(2):19–21. *[Crossref][PubMed][Google Scholar]*

5. Dole VA. Rasaratnasamuccaya. 3rd ed. Ch. *5, Verses 9–11. Varanasi: Chowkhamba Sanskrit Series Office; 2015. p. 166–7 [Crossref][PubMed] [Google Scholar]*

6. Pandit KS, editor. Rasatarangini of Pranacharya S. 8th ed. Ch. *15, Verse 69. Delhi: Motilal Banarsidass; 2014. p. 375 [Crossref][PubMed][Google Scholar]*

7. Nishteswar K, Vidyanath R. Sahasrayogam. Reprint ed. Taila Prakarana, 40. *Varanasi: Chowkhamba Sanskrit Series Office; 2017. p. 135 [Crossref][PubMed][Google Scholar]*

8. Nishteswar K, Vidyanath R. Sahasrayogam. Reprint ed. Taila Prakarana, 13. *Varanasi: Chowkhamba Sanskrit Series Office; 2017. p. 117 [Crossref][PubMed][Google Scholar]*

9. Venkatesh P, Ashivini. Effect of Dashmoola Siddha Ksheera Paka Kati Basti in Katishoola – A single case study. J Ayu Int Med Sci. 2022;7(4):127–32. *[Crossref][PubMed][Google Scholar]*

10. Murthy KRS, editor. Vagbhata's Astanga Hrdayam. Vol 3, Uttarasthana, Ch. 1, Verse 45. *Varanasi: Chaukamba Krishnadas Academy; 2018 [Crossref][PubMed][Google Scholar]*

11. Mishra SN, editor. Bhaisajya Ratnavali of Kaviraj Govind Das Sen. Vajikarana Adhikara. Varanasi: Chaukhamba Surbharati Prakashan; [no year provided]. *p. 117 [Crossref][PubMed][Google Scholar]*