

AYURVEDIC UNDERSTANDING AND MANAGEMENT OF DOWN SYNDROME - A CASE REPORT

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ABSTRACT

Down Syndrome or Trisomy 21 is the most common autosomal trisomy which accounts for more than one third of infants who are born with a chromosomal abnormality. Down Syndrome occurs more often in offspring of mothers conceiving at older age. Down Syndrome or Mongolism is the most common among the well-recognized causes of mental retardation. Incidence in India is 2.2 per 1,000 live births which is comparatively higher than the average overall figure of 1 in 600 for all races. Diagnosis is most likely made in the new born period itself. Children with Down Syndrome usually has both intellectual disability as well as physical retardation along with a flat facial profile and an upward slanting of eyes and epicanthic folds. These children suffer from recurrent respiratory infections and associated congenital heart disease is common in them. Most prominent feature is mental subnormality. These children will be there in the IQ range of 25-50. A 9 year old male child was brought to SDM College of Ayurveda and Hospital, Hassan with a confirmed diagnosis of Down Syndrome by his parents with complaints of delay in attaining speech and lack of clarity in speech along with less socialization and reduced intelligence level. In Ayurvedic perspective this condition can be understood as a disease occurring due to *Beeja Beejabhaga* and *Beejabhagavayava dushti*. After proper examination and clinical evaluation, treatment was initiated. Judicial administration of *Medhya*, *Balya*, *Avaranahara* and *Rasayana oushadha* also helped in improving the condition of the patient. Overall, a significant improvement was noted.

Keywords: Down Syndrome, *Beeja Beejabhaga Beejabhagavayava dushti*, *Medhya*, *Rasayana*.

INTRODUCTION

Down Syndrome was first described by Langdon Down in 1866. However, the chromosomal defect was unidentified till 1959. In that year, Lejeune and his associates found that patients with this condition have 47 chromosomes instead of the normal 46¹. Down

Syndrome or Trisomy 21 is the most common autosomal trisomy which accounts for more than one third of infants who are born with a chromosomal abnormality². Down Syndrome occurs more often in offspring of mothers conceiving at older age. The risk in

new born is 1:1550 if maternal age is 15-29 years, 1:800 at 30-34 years, 1:270 at 35-39 years, 1:100 at 40-44 years and 1:50 after 45 years of age³. It is considered as the most common abnormality of chromosomal number. It occurs in 1 of every 1000 births. Most cases (92.5%) are due to nondisjunction; in 68%, the non-disjunctional event occurs in maternal meiosis phase I. As a result of this nondisjunction, there will be three copies of chromosome 21 (Trisomy 21). By using standard cytogenic nomenclature, it can be designated as 47, XX, +21 or 47, XY, +21. In 4.5% cases, this extra chromosome is part of a Robertsonian translocation, which occurs when the long arms (q) of two acrocentric chromosomes (numbers 13,14,15,21 or 22) fuse at the centromeres and the short arms (p) containing copies of ribosomal RNA are lost⁴. In about 1-2% of children with Down Syndrome, mosaicism occurs. They have two populations of cells; one with trisomy 21 and one with a normal chromosome complement.

Down Syndrome is the first chromosomal disorder to have been clinically defined. Recent studies suggest that paternal age may also be related to the incidence of Down Syndrome⁵. Down Syndrome or Mongolism is the most common among the well-recognized causes of mental retardation. Incidence in India is 2.2 per 1,000 live births which is comparatively higher than the average overall figure of 1 in 600 for all races⁶. Diagnosis is most likely made in the new born period itself. They used to have a normal birth weight and length as infants. The severe hypotonia troubles them with feeding problems and reduced activity⁷. As the child grows, hypotonia gradually diminishes. Most prominent feature is mental subnormality. These children will be there in the IQ range of 25-50.

Children with Down Syndrome usually has both intellectual disability as well as physical retardation along with a flat facial profile and an upward slanting of eyes and epicanthic folds. Their nose is comparatively small with a flat nasal bridge. Mouth is typical with a narrow short palate, small teeth and furrowed protruding tongue (scrotal tongue). Presence of hypotonia is significant. Skull is small and brachycephalic with a flat occiput. Ears are small. Their hands are short and

broad. Clinodactyly (hypoplasia of middle phalanx of fifth finger) and single transverse palmar crease (Simian crease) are typical. A wide gap between the first and second toe known as Sandle gap is yet another peculiarity⁸. Occasionally, brushfield spots (small whitish spots near the periphery of the iris) can be seen. Their neck is short and broad. Most often their head seems to be almost resting on trunk. Hairline is usually low. These children suffer from recurrent respiratory infections. Associated Congenital heart disease (ASD or VSD or TOF) is common in them. They are 10 times more prone to the development of leukemia than the normal population⁹. They may have a delay in attaining developmental milestones appropriate for age and even sensorineural deafness. Hypothyroidism is quite common (25-40%) in them. Approximately 10% newborns with Down Syndrome have gastrointestinal tract anomalies. The three most common defects are duodenal atresia, annular pancreas and imperforate anus.

The Principles of management includes giving early stimulation, physiotherapy, speech therapy, adequate nutritional supplementation, education, visual and hearing assessment and aids¹⁰. Whenever required, associated problems need to be properly treated. Along with that, occupational therapy, specific exercises that can improve the gross and fine motor skills, surgery in case of moderate to severe abnormalities of heart is also recommended¹¹. Social performance will be usually achieved beyond what was expected for mental age. They behave as happy children who love mimicry, friendly and music lovers. Helping the child to bring the best out of his limited abilities is the target. Counselling to be given to the parents is very much crucial. As the parents need to be strong enough and well prepared to accept the reality and are the main people who need to help these children to do their best in day to day life. Regarding the risk of recurrence especially, women of 35 years of age or less who have a child with trisomy 21 have a 1 % risk of having another, which is significantly greater than the general population. The chief reason for early mortality in such children is congenital heart disease. Almost 50% children with cardiac anomalies die in infancy.

Chronic rhinitis, conjunctivitis and periodontal disease are common. Lower respiratory infections possess a threat to their life. Even hematological malignancies are yet another cause for the mortality. These children usually lead a partly independent life of 50-55 years. As they become older, there is an increased risk of associated ailments. All these associated complaints also need to be addressed properly so as to offer them a better life.

Ayurveda explains in detail about the defects in *Beeja* (seeds), *Aatma karma* (actions associated with the soul), *Aashaya* (uterus), *Kaala dosha* (time), *Matustataahara vihara dosha* (food and regimen of mother), results in the vitiation of *Doshas* and resulting in impairment of *Samsthaana* (shape), *Varna* (colour) and *Indriya* (sensory as well as motor organs of offspring). Main one among them is the vitiation of *Beeja* (sperm or ovum) that causes damage of genetic material¹². One human being is the product of another human being, but a progeny of a dull human being is not always dull. If the part of seed (sperm or ovum) which is responsible for the formation of a particular organ gets vitiated, that will result in the vitiation of the respective organ. If it is not vitiated, then there would be no vitiation of the respective organ. Hence both the possibilities exist¹³. Vitiating *Doshas* afflicts the *Beeja* (can be understood as chromosome in modern genetics) or the *Beejabhaga* (can be understood as gene in modern genetics) by which the *Avayavaas* (corresponding organs) derived from these two gets *Vikrita* (deformed)¹⁴.

Many studies have revealed that pharmacotherapy may be helpful in ameliorating or lessening the intellectual impairment in animals and human beings. Strengthening of the intellect chiefly depends on factors like status of *Agni*, status of *Prana* and *Udana vata* and the status of auditory, visual and proprioceptive *Indriyas*¹⁵.

Budhi nirodha (Mental restriction) is mainly due to excess *Tamo guna*¹⁶. *Mana* and *Vayu* having similar functions work together like water in milk. By activating *Vayu*, *Mana* gets activated. If one gets *Kshaya*, it leads to *Kshaya* of other. Counselling and education along with giving special attention to such children

helps in improving *Dhee*, *Dhairya* and *Aatma vignaana*¹⁷. Down syndrome can be even considered under *Adidaivika Vyadhi*, which occurs because of those factors which are beyond human access, influence or control. The disease caused due to bad deeds done in present or past life is giving its effect back in the form of disease or suffering¹⁸. Some, even consider Down Syndrome under *Adibala Pravritta Vyadhi* as well. If both *Shukra* and *Shonitha* becomes *Nashta*, there is no *Praja*, but if any part of either *Shukra* or *Shonitha* (*Beeja bhaga*) is diseased or *Roga grastha* then there will be *Praja* but with some pathological manifestations. This can be again divided into two-*Matruja* (inherited from female or mother to progeny) and *Pitruja* (inherited from male or father to progeny)¹⁹.

CASE HISTORY:

1. A 9 year old male child who is a known case of Down Syndrome was brought to SDM College of Ayurveda and Hospital, Hassan by his parents with complaints of delay in attaining speech and lack of clarity in speech along with less socialization and reduced intelligence level.
2. This child was born by Full term normal vaginal delivery in a hospital in Karnataka. Baby cried immediately after birth. No any obvious external congenital anomalies were present. No any history of NICU Stay. Breast feeding initiated immediately after birth. No any history of consanguineous marriage.
3. Few days after birth, somebody has told them that there is some difference in the facial appearance of this child when compared with other new born babies. But they have not taken it seriously. Later, they have noticed by about 6 months of age, that the child has some facial dysmorphism. There was a slight delay in attainment of the developmental milestones as per age. Then, especially when the child has not developed speech other than cooing and jargons by even 2 years of age, they have decided to consult a Pediatrician in Mysuru.
4. Proper evaluation and diagnosis was made in a hospital in Mysuru that this child is a case of

Down Syndrome. Parents were given a detailed information regarding this condition and counseling was also done by them. They were asked to observe still, if there is any delay in attainment of developmental milestones from then.

5. This child was having recurrent episodes of cold and cough since childhood, which has comparatively reduced since a year. There was no any Congenital heart Disease which got confirmed after the series of evaluations in Mysuru hospital.
6. As he grown up, parents have sent him to a nearby school and he was not following what was dealt by the teachers in class. He loves to play with friends and loves music. But he cannot speak much. Hardly, he speaks monosyllables Aa, Ee, Uu etc and smiles often and communicate most of the things by action. He has slight difficulty in following commands given by anybody other than parents. He has a typical walking pattern and remains silent most of the time.
7. He uses only one bisyllable that is AMMA, which he started by the age of 5 years. No any other bisyllables he speaks. He even responds to his father with a smile or by catching his hand or shirt. He was presenting with lack of concentration and not understanding commands and things which needs proper efforts.
8. Even though, Speech therapy was initiated few years before, for him and continued for a period of 2 months, but they could not continue it due to familial reasons and lack of cooperation by the child. Parents have heard from some health workers that many such children are receiving treatments in this hospital, they have decided to bring him to SDM College of Ayurveda and Hospital, Hassan for better management. After a detailed interrogation with parents and a thorough examination and proper evaluation, the child was admitted in SDM Ayurveda Hospital.

ON EXAMINATION:

Table 1: Assessment of general condition of the child:

Bowel	Regular
Appetite	Good
Micturition	Regular
Sleep	Sound

Table 2: Physical examination (Head to Toe) of the child:

Face	Typical-Mongolian facies
Eyes	Upward slanting of eyes and epicanthic folds present
Nose	Comparatively small nose with flat nasal bridge
Mouth	Protruding tongue
Neck	Short neck and low hair line
Hands	Short and broad
Feet	Wide gap between the first and second toe present

Table 3: Systemic examination

Cardiovascular System (CVS)	S1 S2 heard, no murmurs
Respiratory System (RS)	Normal Vesicular breathe sounds heard, Bilateral air entry +
Gastro intestinal System (GI)	P/A-Soft, non tender.
Vision	No any abnormality detected
Hearing	No any abnormality detected

TREATMENTS GIVEN:

First Sitting (10 Days):

External treatments:

1. Sarvanga Abhyanga with Mahanarayana thaila, followed by Nadi Sweda
2. Matra Basthi with Samvardhana Ghrita (40ml)
3. Prathimarsha Nasya with Anu Thaila 2 drops/2 drops (BD)
4. Pradhamana Nasya with Vakshudhikara churna
5. Shirothalam with Brahmi thaila + (Powder combination of Medhya dravyas)-Aswa-gandha+Vacha+Yashtimadhu+Shankhapushpi+Jatamansi+Mandukaparni
6. Asyaprathisarana with Vakshudhikara churna+honey.

Internal Medications:

1. Saraswatharishta (7.5ml BD with equal amount water) A/F
2. Medhya Vati (1 BD) A/F with honey
3. Kushmanda Swarasa (30ml BD) A/F
4. Hingwashtaka Churna (1tsp TID with food)
5. Kumarabharana Rasa (1 Tab OD with honey) B/F

Second Sitting (10 days):

External treatments:

1. Sarvanga Abhyanga with Mahanarayana thaila, followed by Nadi Sweda
2. Matra Basthi with Samvardhana Ghrita (40ml) for first 2 days.
3. Yoga Basthi: Niruha Basthi with Bala Dashamoola Kashaya Basthi (270 ml) Anuvasana Basthi with Samvardhana Ghrita (40 ml)+ Brahmi thaila (30 ml)-Total: 70ml
4. Prathimarsha Nasya with Anu Thaila 2 drops/2 drops (BD)
5. Pradhamana Nasya with Vakshudhikara churna
6. Shirothalam with Brahmi thaila + (Powder combination of Medhya dravyas)-Aswa-gandha+Vacha+Yashtimadhu+Shankhapushpi+Jatamansi+Mandukaparni
7. Asyaprathisarana with Vakshudhikara churna+honey.

8. As per Yoga Basthi plan, Anuvasana Basthi was given on 3rd, 5th, 7th, 9th & 10th day and Niruha Basthi was given on 4th, 6th and 8th day during the course of treatment of 10 days.

Internal Medications:

1. Saraswatharishta (7.5ml BD with equal amount water) A/F
2. Medhya Vati (1 BD) A/F with honey
3. Kushmanda Swarasa (30ml BD) A/F
4. Brahmi Ghrita (1tsp BD with honey) B/F
5. Hingwashtaka Churna (1tsp TID with food)

ADVISE AT THE TIME OF DISCHARGE:

1. Asyaprathisarana with Vakshudhikara churna with honey
2. Prathimarsha Nasya with Anu thaila 2 drops/2 drops (BD)
3. Hingwashtaka Churna (1tsp TID with food)
4. Saraswatharishta (7.5ml BD with equal amount water) A/F
5. Kumarabharana Rasa (1 Tab OD with honey) B/F
6. Medhya Vati (1 Tab BD with honey) A/F
7. Brahmi Ghrita (1tsp BD with honey) B/F

OUTCOME OF THE TREATMENTS:

Patient and Care taker's Feedback, after the first course of treatment:

1. General health status has improved.
2. The child started observing things better than before. Concentration has improved.
3. Started demanding for things by showing and trying to express what it is.
4. Appears more happy and comfortable with the surrounding environment and started interacting with other children and started showing more interest in play activities.
5. Appetite has improved.

Clinician assessed outcomes:

1. Child's overall health has improved. He was looking silent, moody and dull during admission. After discharge, he was more enthusiastic, happy and cooperative.
2. His observation skills has improved and it was noted that he was concentrating on picture books, toys, birds and towards various people around him especially what they talk.

3. He started asking for toys, water, food, bag, clothes better than previous by trying to tell the initial letters.
4. Child appeared more comfortable with friends, strangers and was dedicating himself actively in playing with other children.
5. Overall appetite has improved and digestive capacity has also improved.

Patient and Care taker's Feedback, after the second course of treatment:

1. Started responding to questions when asked and trying to answer it properly.
2. Started speaking few more bisyllables like Appa, Chinni etc.
3. Initially he was not happy with his parents taking any child in their hand and playing or talking to them. He used to get angry for that.
4. Started understanding "No" when told by parents for something which should not be done.
5. Child started showing more interest in food and he asks when feels hungry.
6. General health has improved a lot. Earlier, since childhood he was having recurrent respiratory infections. Now, for the past 1 month there was no any episode.

Clinician assessed outcomes:

1. When his name was called, he used to smile and show typical expression in face.
2. Child started telling Appa, started calling babies in nearby bed as Chinni.
3. Child is better and started not reacting badly to parents when they take any kids and play. Even he started keeping small kids in his lap and trying to love them.
4. Child started responding to "No" by parents and immediately stops what he is doing.
5. He used to touch our stethoscope and make us to check him and he even started showing where exactly it should be kept.
6. Appetite has very well improved.
7. General health has also improved. No any attacks of cold and cough for almost a month.
8. Child's understanding and concentration has improved.

DISCUSSION

Beeja is a part of ovum or sperm which is responsible for the development of a particular organ. The nearest term in the modern perspective to which it can be correlated is chromosome. *Beejabhaga* is a part of *Beeja*, which can be correlated to gene in modern parlance. Whereas, *Beejabhagavayava* can be understood as DNA. According to Ayurveda classics, these type of diseases occurs not only due to *Beeja*, *Beejabhaga*, *Beejabhagavayavadushti*, but also due to *Mithyahara vihara* by mother and *Garbhini Avamanana* as well. When there is lack of proper intelligence, delayed speech development, these type of diseases can also be categorized under *Samvardhana Vikara*, in which the normal growth and development in a child gets affected. *Tridoshas* plays a key role in the proper functioning of both *Sharira* and *Manas*. When *Prana vayu* gets afflicted, it affects the normal functioning of *Budhi*, *Indriya* and *Chitta*. In the case of *Udana vayu*, when afflicted, results in the abnormal functioning of *Vak pravritti*, *Prayatna*, *Bala*, *Varna* and *Smriti*. *Vyana vayu* is related to various thought processes. *Samana Vayu* has the capacity to normalize all the other *Vayu*. When *Sadhaka Pitta* gets afflicted, it results in abnormalcy in achieving the intended objects with *Budhi* and *Medha*. *Tarpaka Kapha* situated in the head, when afflicted, results in improper functioning of all the *Indriyas*. In *Pranaavrita Vyana vayu*, there will be *Sarvendriyanam shunyatva*. Whereas in *Pranaavrita Samana Vayu*, there will be *Jada*, *Gadgada*. In *Kaphaavrita Udana Vayu*, there will be *Vakswaragraha*. Treatment was planned in order to bring equilibrium to the status of *Tridosha-Vata*, *Pitta* and *Kapha* and also, to help in bringing normal functions of all these *Prana*, *Udana*, *Samana*, *Vyana vayu*, *Sadhaka pitta* and *Tarpaka Kapha*. *Avaranahara chikitsa* also brings better results. Judicial administration of *Medhya*, *Balya* and *Rasayana oushadha* also helped in improving the condition of the patient. *Snehana* in the form of *Sarvanga Abhyanga*, which is a *Bahya sehana karma* was given as it has property of *Sneho anilam hanti* or *Vatahara* property and brings back *Doshas* from *Shakha* to *Koshta*. *Mahanarayana*

thaila was opted for *Abhyanga* as it is *Vatahara*, *Ayushya* and *Balavarnakrit*. *Swedana* with *Nadi sweda*, which is one among the *Ashta Sweda* of *Kashyapa*. In general, *Swedana* helps in *Agnideepthi*, *Twak prasada*, *Bhaktashradha* and *Srothonirmalata*. Initially, *Matra Basthi* which is a type of *Sneha basthi* specifically indicated in *Bala* and *Sukumara* was chosen. *Samvardhana ghruta* helps in *Nirvyadhi vardhana* and helps *Muka*, *Asruti* and *Jada* to perform their normal functions. *Murdhni thaila* were judiciously administered which targets head, which is having a special importance among all the body parts, hence known as *Uttamanga*. *Shirothalam* was done with a combination of *Medhya dravya* like *Brahmi* which is *Medhya Rasayana* and promotes speech, *Aswagandha* which is *Balya* and *Rasayana*, *Shankhapushpi* which is *Medhya* and *Rasayana*, *Yashtimadhu* which is *Rasayana*, well known brain tonic and improves speech, *Vacha* which helps in improving intellect and speech, *Jatamansi* which is *Medhya*, along with *Brahmi thaila* which is *Tridosahara*, *Medhya*, *Smritiprada* and *Chittaprasadaka*. *Brahmi ghruta* was given internally as it has *Vak swara smriti medhakrit* and *Vakshudhi* action. *Saraswatharishta* which helps in *Vakshudhi*, *Dhairya* and *Smriti labdi* would have also helped the child. In the second sitting *Yoga basthi* was administered. *Basthi* with qualities like *Pushtikara*, *Medhakrit*, *Budhindriya samprasada*, *Swapnavritti* and *Harsha*. *Niruha Basthi* is a *Shodhana* type of *Basthi* helped in removing the vitiated *Dosha*. *Anuvasana basthi* helped in correcting *Vata dosha* with its target action in *Pakwashaya* which is the *Visesha sthana*. *Nasya* is specifically indicated in Speech disorders like *Vak graha*. *Nasya* helps in attaining *Indriya shudhi*, *Sukha Swapna* and *Manasukha*. *Pradhamana Nasya* was given which is specifically indicated in *Chetovikara* and *Utkata dosha*. *Prathimarsha Nasya* is advised to be performed in *Bala* and *Sukhatma*. *Kumarabharana Rasa* internally helps in improving the immunity and also to improve intellect. *Medhya vati* was also for *Medhya rasayana* action. All these therapies together can be considered as an Ayurveda treatment package which was judiciously administered which helped in improving the condition of the child.

CONCLUSION

Down Syndrome is a genetic disorder. It occurs as a result of chromosomal abnormality and one of the major reasons for mental retardation. The Principles followed for its management includes giving early stimulation, physiotherapy, speech therapy, occupational therapy, adequate nutritional supplementation, education, visual and hearing assessment and aids. Associated problems also need to be treated. In Ayurvedic Perspective, this disease condition can be understood as that caused by *Beeja*, *Beejabhaga* and *Beejabhagavayava dushti*. It is categorized under *Adidaivika* and even *Adibalapravritta vyadhi*. Judicial application of *Medhya*, *Balya*, *Avaranahara* and *Rasayana oushadha* also helped in improving the condition of the patient. Down Syndrome cannot be cured completely. The quality and standard of life of such children can be improved with Ayurveda line of management. Intelligence level can be improved to an extent and any developmental delay can also be managed. Even associated ailments can be managed by improving immunity and overall general health of the child.

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