Spondyloepiphyseal dysplasia tarda (SEDT) is an X-linked hereditary skeletal disorder however, it may be transmitted as autosomal recessive or autosomal trait [1]. Tarda indicates the manifestation of disorder is not present at birth but appear in childhood typically between age 6 and 10 years. Prevalence of SEDT is not established but according to one study it was estimated to be at least 1.7 in 1,000,000 people without any ethnic predisposition [1,2]. Epiphyseal involvement is primarily in the shoulders, hips and knees. This involvement is symmetrical bilaterally. SEDT is characterized by disproportionately short stature with short trunk, barrel-shaped chest and arm span significantly greater than height. At birth, affected males are normal in length and body proportion usually about 0.8, Arm span typically exceeds height by 10–20 cm. Final adult height is typically 137–163 cm. Characteristic radiographic findings and diagnostic markers include: platyspondylly with characteristic superior and inferior humping seen on lateral view, narrow disc spaces, scoliosis, and coxa vara and evidence of osteoarthritis beginning in young adulthood. Only limited medical and surgical management is available in modern medicine. A 15 years old male suffering from SEDT and diagnosed as Vata vyadhi was treated with Panchakarma therapy and selected Ayurvedic oral medicines. Ayurvedic treatment was directed to ameliorate the orthopaedic clinical conditions in this case. Panchakarma procedures such as Shalishastika pinda svedana for a month and Mustadi yapana basti for 16 days were given along with oral Ayurvedic medicines. Same Panchakarma procedures were repeated after an interval of 2 months. A combination of Ayurvedic oral medicines such as Trayodashanga guggulu-500 mg twice a day, Dashmool kvatha (decoction of roots of 10 herbs) 40 ml twice a day, Eranda paka 10 g twice a day, Shiva gutika-500 mg twice a day and Dashmoolarishta-20 ml (with equal water) twice a day were prescribed. Eight scales based Medical outcome study (MOS) – 36 item short form – health surveys was assessed for outcome which shows good improvement. Kyphosis, scoliosis and pain were moderately reduced. Clinical experience of this case indicates that Ayurvedic herbs along with Panchakarma can play a major role in the management of hereditary disorder SEDT.
with medicated milk] were adopted as Panchakarma procedures and Trayodashanga guggulu, Dashmool kwatha ( decoction of roots of 10 herbs), Eranda paka, Shiva gudika and Dashmoolarista as selected Ayurvedic oral medicines. Signs and symptoms of SEDT at various stages are similar to be various Nanatamaj Vata Vyadhi (specific diseases due to only Vata dosha) as described in Ayurveda. Thus the patient in this case report was considered to suffer from Vata Vyadhi as Ayurvedic diagnosis. The clinical observations of this case is important as there is no report on Ayurveda management of SEDT.

2. Case report

A 15 yrs old Indian, male patient was consulted in Out-Patient Department of National Institute of Ayurveda, Jaipur with complaints of short stature, progressive deformity of hands, elbows and knees, lower backache, stiffness, pain in all joints, difficulty in walking and squatting and difficulty in breathing on exertion.

Patient was asymptomatic till 5 years of age then there was gradual increase of difficulty in walking. Pain and swelling in fingers (metacarpophalangeal joints) were noticed at the age of 6 years. By the age of 7 years thinning of legs were prominent. Severe walking disturbance and contracture in fingers had developed at the age of 8 years. Chest deformity was prominent till 10 years of age. Patient was repeatedly consulted in S.M.S. medical college Jaipur, Rajasthan from 5 to 7 years of age. Patient was further consulted in A.I.I.M.S. New Delhi in the year 2014, where gene mapping was done and he was diagnosed for SEDT. Patient had received non-steroidal anti-inflammatory medicines and various analgesics but his complaints did not respond to these treatments. Patient’s neuromotor development was normal. Family history revealed that his cousin sister was also affected from the same manifestation. This is a rare finding however a case of a female with Turner syndrome, affecting from SEDT has been reported [4]. Patient was admitted in the Panchakarma ward of National Institute of Ayurveda, Jaipur on May 21, 2015 for administration of Panchakarma procedures.

On physical examination patient was found to be anxious with disturbed sleep, Vishmagni (unstable digestive functions), bowel constipated, burning micturation with reduced frequency, voice clear, roughness in skin on touch and lean body built. The patient was in Niramavastha [stage of disease without Ama (~undigested food)] with apparently normal appetite. There was no abnormal coating on tongue. Patient had Vatapitta prakriti with Avara sara (sub optimal body tissue), Avara samhanana (sub optimal body built), Vishama pramana (unequal body proportion), Avara satmya (sub optimal homologation), Madhyayam satva ( medium mental strength), Avara vyayamshakti (sub optimal capability to carry on physical activities), Madhyam aharashakti and Jaranshakti (medium food intake and digestive power). Asthivaha srotodusti (pathology in bone) and Majavaaha srotodusti (pathology in bone marrow) were more prominent. Weight was 22.1 kg, height was 128 cm, arm span was 140 cm and head circumference and distance between two eyes were normal. Higher function, mental state and speech were normal. Neurological, skin, cardiorespiratory and genitourinary system examination were normal. Audiometric results were normal. Bilateral peripheral cataract was found on slit-lamp examination of eye. The internal and external rotation of hips, the flexion and abduction of shoulders, the extension of elbows and knees were limited. Ankles and wrist were ankylosed. 5th Distal Interphalangeal joint and 2nd to 4th proximal phalangeal joints were swollen with flexion deformity [Fig. 1] Lumbar and cervical spine extensions were restricted with pain. Feet were flat and toes were abnormally long. Neck was short and thoracic kyphosis, scoliosis and barrel shaped chest were found [Fig. 2] In laboratory analysis complete blood count, lipid profile, routine urine analysis, liver and kidney functions were normal.
of hip region), **Pristhagraha** (stiffness of back), **Pasravamarda** (pain in coastal region), **Timira** (early cataract) **Ucchaisruti** and **Badhirya** (different level of deafness), **Janubheda** and **Januvishlesha** (osteo-arthritis of knee), **Vatakhudata** and **Padabhransa** (structural deformity in sole) **Gulfagraha** (stiffness in ankle), **Vakshauprodha** (restrictive pulmonary movement). Thus the patient suffered from a group of several **Nanatamaj vata vyadhi**.

**Mukhasosha** (coarse facial appearance), **Akshivyodash** (wide set eyes), **Bhruvyodasha** (wide nose bridge), **Akshibheda** (strabismus), **Mukatvam** and **Vakasanga** (speech impairment), **Dantabheda** and **Dantasaithilya** (dentinogenesis imperfect) are the other symptoms which may be prominent in other variant of SEDT [5]. Considering all these pathogenesis and manifestation, **Vata vyadhi** was considered as Ayurvedic diagnosis for the case. Anterior beaking of vertebrae with generalized platyspondyly and marked hip and knee metaphyseal lesions are characteristics of SEDT that was present in the patient. Arm span of the patient was 12 cm more than height in the case. This is described in Ayurveda as **Visham pramana**, also a characteristic of **Vata vyadhi**.

Spondyloepiphyseal dysplasia congenital (SEDC), Morquio syndrome, multiple epiphyseal dysplasia (MED) and **Vatarakta** are the differential diagnosis for the case. SEDC is an autosomal dominant disorder which equally affects males and females. This disorder is often detectable at birth. Morquio syndrome is usually detectable at 18 months of age. Excretion of excessive mucopolysacharides in the urine is characteristic of Moriquio disease. MED is usually dominantly inherited and equally affects males and females. As these symptoms were first noted at the age of 5 years so this was considered a case of SEDT and from Ayurvedic point of view **Vata vyadhi** was considered as Ayurvedic diagnosis. The differential Ayurvedic diagnosis for the case was **Vatarakta** (various diseases of rheumatic spectrum). There was no **Rakta Dusti** (vitiation of blood) and also **Purvaroopa** (prodromal symptoms) of **Vatarakta** were not observed hence diagnosis of **Vatarakta** was ruled out.

## 4. Management

Patient was suffering from SEDT. **Vata vyadhi** was considered as Ayurvedic diagnosis. Considering the chronic nature of disease, the Ayurvedic management was directed to ameliorate the various symptoms and complications of disease. The general management of **Vata vyadhi** as **Snehana**, **Svedana**, **Mridu virechana** (mild purgation) and **Basti** (medicated enema) was applied in this case [6]. **Basti** is indicated as best treatment for any **Nanatamaj vata vyadhi** [7]. All of these principles were adopted in the management of the patient.

For the first 3 days **Mridu virechan** was administered with castor oil in the dose of 20 ml with milk. From 4th day **Shalishastika pinda svedana** for a month along with **M. yapana basti** for 16 days were administered [Table 1]. **Abhyanga** (massage) with **Asvagandha taila** (oil) was the integral part of the **Shalishastika pinda svedana**. These procedures were repeated after an interval of 2 months. Oral Ayurvedic medicines such as **Trayodashanga guggulu-500 mg twice a day**, **Dashmool kwatha** (decoction of roots of 10 herbs) 40 ml twice a day, **Eranda paka 10 g twice a day**, **Shiva gutika -500 mg twice a day** and **Dashmoolarista-20 ml with equal water twice a day** were also prescribed along with **Panchakarma** procedures during this period [Table 2]. Clinical outcome was assessed at completion of first round of **Panchakarma** procedures and at the end of second round of **Panchakarma** procedures.

## 5. Timeline

A detail of the case study and follow up is given in [Table 3].
was increased by 1.9 kg. This was due to (Wild.) Hook. F. 

Table 3

6. Outcome measures and follow up

Good relief was noted by the patient [Table 4]. Walking and lung capacity improved after a month of treatment. Kyphosis, scoliosis and pain were significantly reduced. Weight of patient was increased by 1.9 kg. This was due to Brihmana (nourishing) and Srotosodhana (purification of channel) effect of therapy. Eight scales based on medical outcome study (MOS)-36 item short form-health surveys were assessed for outcome which shows improvement [8]. No concurrent allopathic medication was administered during this period. Patient was discharged on 8th Oct 2015 with instruction to continue same Ayurvedic medicines for longer duration and repetition of Panchakarma procedures after 2 months. Outcome measure was assessed on baseline, after completion of first Panchakarma schedule and at the completion of second Panchakarma schedule.

Table 3

Timeline.

| Year      | Events                                                                 |
|-----------|------------------------------------------------------------------------|
| 2005      | - Onset of problem in walking, was consulted in J.K.Loan hospital Jaipur, Rajasthan |
| 2006      | - Starting of contracture in fingers, was consulted in S.M.S. medical college, Jaipur, Rajasthan. |
| 2007      | - Thinning of leg were prominent.                                      |
| 2008      | - Severe walking disturbance and contracture in fingers were developed. |
| 2010      | - Chest was deformed.                                                  |
| 2014      | - Was further consulted in a reputed tertiary care hospital and was diagnosed for SEDT. Gene mapping was done. M.R.I. screening of whole spine that was conducted on May 21, 2014 and digital X-rays revealed osteopenia, flexion deformity of 2–4th distal phalangeal joint and 5th distal Interphalangeal joints, metaphyseal sclerosis of femur and tibia (knee), irregular acetalbumin, coxa vara, anterior beaking of vertebral with generalized platyspondyly and disc desication from C3/4 to D11/12 and L4/5, L5/S1 levels. |
| 21/05/2015| - Patient was admitted in O.P.D. of National Institute of Ayurveda for progressive skeleton deformity. Clinical assessment for disease based on Medical outcome study (MOS)-36 item short form-health survey was done. [Table 4] |
| 22/05/2015-24/05/2015 | - Castor oil was given at night for mild purgation. |
| 25/05/2015–23/06/2015 | - Shalishastika pinda svedana for one month along with Mustadi yapana basti for 16 days were adopted as Panchakarma procedures. Ayurvedic oral medicines such as Trivadashangha guggulu-500 mg twice in a day, Dashmool kwatha (decoction of roots of 10 herbs) 40 ml twice a day, Eranda paka 10 gm twice a day, Shiva gutika-500 mg twice a day and Dashmoolarista-20 ml with equal water twice a day were also prescribed along with these procedures. Ayurvedic oral medication is continued till date. |
| 01/07/2015 | - Patient was discharged and assessment for clinical improvement was done. [Table 4] |
| 21/08/2015 | - Patient was readmitted in O.P.D. for further Panchakarma procedures. |
| 23/08/2015–23/09/2015 | - Same Panchakarma procedures and oral medication were repeated for same duration. |
| 08/10/2015 | - Patient was discharged and assessment for clinical improvement was done. [Table 4] |
7. Discussion

SEDT is a rare hereditary disorder due to abnormal synthesis of sedlin protein. This abnormal synthesis leads to manifestation of disease. Any abnormal manifestation may be due to Vikrita Vata (vitiated Vata). Vitiated vata can also lead to hereditary disorders [9]. Vata is vitiuated due to several etiological factors, Margavarana (obstruction in natural course of vata such as normal distribution, synthesis of tissues elements etc.) and Dhatukshaya (~depletion of body tissue). The vitiated vata also leads to Margavarana and Dhatukshaya in vicious cycle and lead to manifestation of SEDT. The patient was treated on the line of management of Vata vyadhi. Yapana Basti, Rasayana, Shiilajatu and Guggulu are indicated for N. vata, Avrita vata and chronic Vata vyadhi [10]. As patient was in the adolescent age mridu basti in the form of Yapana basti and mridu snehana and svedana in the form of Shalishatika pinda svedana were prescribed. Basti can break pathogenesis of Vata vyadhi by removing Margavarodha by purification of channels and Dhatukshaya by its Brihmana (~nourishing) property. M. yapana basti is indicated for increasing vigour, strength and semen. It is useful in Katishoola (backache), pain in thigh and calf region, abdominal pain, headache, cough and Vatarakta (various diseases of rheumatic spectrum). It also has Rasayana property. Most of Majja and Asthi pradoshaj viKar (disorders of bone and bone marrow) can be managed with M. yapana basti [11]. Slight modification was done in M. yapana basti for this case [12,13]. Trayodashanga guggulu is useful in Snayagatavata (~various tendon and ligament disorders), Asthigatavata (disorders of bone), Majjagatavata (disorders of bone-marrow), Khanjavata (limping disorders), and various Vatic disorders (~neurological, rheumatic and musculoskeletal diseases) [14]. Dashamoola kvatha is having TridoshaNetra property [15]. Shiva gurika is a Rasayana and helpful in Vatarakta, Shiroroga, Mukharoga, Swas roga (dyspnoea), Neteraroga etc. [16] Eranda paka is indicated in Vata vyadhi, pashavadha (paralysis), Amavata, Kativata, Panguyla, Shoolaa (abdominal colic), Vibandha (constipation), Kasaa (cough) etc. [17] Dashmoolarista is effective in Vata vyadhi, Mutrakricha (various urological disorders), Mandagni (diminished digestive power), Dhatukshinata (depletion of body tissue) etc. [18] Thus these combinations are useful to manage the manifestation of disease in this case.

The case study shows that such a hereditary musculoskeletal disease can be managed with Ayurvedic treatment. In the present case report, patient started Ayurvedic treatment in the advanced stage of disease. Considering the nature of SEDT, quality of life of the patient can be improved and complication of disease can be reduced with Ayurvedic management. It may be possible to limit the manifestation of disease if the patient approaches in early stages of disease.

8. Conclusion

Ayurvedic management including oral Ayurvedic medicines and Shalishastika pinda Svedana and M. yapana basti was helpful in treating the patient of SEDT. The observations and experiences of Ayurveda management of SEDT may be useful for its treatment and research.

Patient consent

Written permission for publication of this case study had been obtained from the patient.

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Conflict of interest

Nil.

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