Abstract: Patients with skeletal dysplasias face unique challenges because of their physical stature, psychosocial issues and medical complications. Rehabilitation can help them by improving their physical and social independence. Moreover, pseudoachondroplasia has often been mistaken for classical achondroplasia which is the most common type of short limbed dwarfism. Accuracy of diagnosis is important for genetic counseling and patient management. In this report, we present the case of a 37 year old lady who was earlier diagnosed to have achondroplasia. She presented to us with multiple somatic complaints and difficulty in her activities of daily living. While admitted under us, based on the clinical and radiological findings, a diagnosis of pseudoachondroplasia was made. This report describes the features that differentiate the two entities and discusses the rehabilitation challenges and management of these patients.

Keyword: Achondroplasia, Pseudoachondroplasia, Rehabilitation, Skeletal dysplasia. Rehabilitation of persons with skeletal dysplasias helps them by improving social and physical independence. Misbeliefs that such people have limited abilities need to be erased in order to help them maximize their independence. Apart from this, there have been instances where few cases have been misdiagnosed as Achondroplasia instead of Pseudochondroplasia. We present the case of a 37 year old lady who had been diagnosed to have achondroplasia earlier and presented to us with multiple somatic complaints. On detailed evaluation, she was diagnosed to have Pseudoachondroplasia. This report highlights the differences between the two entities and to the best of our knowledge is the first report which addresses the rehabilitation challenges of these patients.

CASE REPORT
A 37 year old lady previously diagnosed to have achondroplasia was admitted with concerns of increased dependence for her activities of daily living (ADL) due to restriction in movements and loss of flexibility, frequent episodes of headache and progressively worsening pain in multiple joints. She was born of normal delivery to non-consanguinous parents with no perinatal complications. There was no history of developmental delay. Neck control was achieved at one month, supported sitting by six months and she started ambulating by eighteen months. After the age of two years, she was noticed to have small stature and waddling gait. Her father also had similar short stature. At a young age, a diagnosis of Congenital Hereditary Achondroplasia was made. She underwent surgical correction of genu varum deformity at 7 years of age. She started having pain in the right wrist, right ankle, the lumbosacral region radiating to bilateral lower limbs and pain in the cervical spine from a very young age. Ten years ago, she started having occipital and frontal headaches associated with nausea and increased sensitivity to light and sound especially in the bilateral patellar subluxation during this period. Worsening pain restrained her from continuing with her daily activities. She was admitted under us for management of her pain and to make her more independent in her activities of daily living. At the time of admission with us, she was walking with a walker. Her walking endurance was about 5m. The main limitations to her walking were pain in the lower back, limbs and joints especially in the ankles, paresthesia over the soles and fatigue on exertion. She was independent in most of her ADL except bathing and perineal care. History of depression and difficulty facing social situations was noted. On clinical examination, there were no facial abnormalities. Head circumference was normal. Height was 119cm with lower segment 50cm and upper segment 69cm. Micromelia with brachydactyly, ulnar deviation of hands, limited extension of elbows, genu vara, mild scoliosis to right, thoracic kyphosis and lumbar lordosis were noted. There was limited range of motion in all her joints. Range of motion of the joints measured using a goniometer is shown in Table 1. She had painful almost habitual subluxation of both patellae left > right. There were no neurologic deficits. Systemic examination was normal.

Table 1: Joint ranges of motion

She was noted to have bilateral ankle instability on weight-bearing. (Figs. 6, 7) Video gait analysis showed bilateral ankles in eversion on weight-bearing. The weight-bearing also showed parameters were within normal limits including inflammatory markers (ESR and CRP). Vitamin
Right Left
Shoulder Flexion 0-100° 0-100°
Shoulder Abduction 0-75° 0-60°
Elbow Flexion 60° - 150° 40° - 150°
Wrist Dorsiflexion 0-30° 0-30°
Wrist Palmar flexion 0-35° 0-35°
Hip Flexion 0-100° 0-100°
Knee Extension 0-110° 0-120°
Ankle Dorsi flexion 0-15° 0-15°
Ankle Plantar flexion 0-55° 0-50°
D and B12 levels were below normal range. Radiographs showed widened horizontal acetabular margins with medial beaking at the femoral neck (fig.1), small, irregular and widened epiphyses (fig.2), malformed vertebral endplates with anterior projections (fig.3), flattened vertebral bodies (platyspondyly) with normal interpedicular distance (fig.4), short metacarpals and phalanges (fig 5). MRI brain and spine done to rule out organic causes of headache, showed evidence of disc with neural foramen compromise from C7 – T1 and no cord compression in the cervical/ lumbar spine. Medical genetics consult was sought to confirm the diagnosis. As per their evaluation, the clinico-radiologic features were suggestive of PSEUDOACHONDROPLASIA. Mutation analysis of gene COMP, genetic counseling and DNA banking were advised.

Figure 1 Widened horizontal acetabular margins, medial beaking at the femoral neck

Figure 2 Small, irregular and widened epiphyses

Figure 3 Malformed vertebral endplates with anterior projections

Figure 4 Platyspondyly with normal interpedicular distance

Figure 5 Short metacarpals and phalanges

Figures 7: Bilateral ankle instability on weight-bearing
Management
The felt needs at admission were improving her ADL independence and ambulation and pain relief. A diagnosis of migraine was confirmed after a Neurology evaluation. She was given analgesics, tricyclic antidepressants, Vitamin D, B12 and Calcium supplementation. She was started on a rehabilitation programme including stretching and strengthening exercises, Vastus Medialis Obliquus (VMO) strengthening for bipatellar subluxation and activities to improve flexibility and decrease lumbosacral pain. She was given gait training with solid ankle foot orthoses bilaterally and walker to provide medio-lateral stability and decrease pain during ambulation (Figs 8,9)* Lifestyle modification, work simplification and joint protection techniques were suggested. Customised aids were provided to improve overhead reach and perineal hygiene. The need for a disciplined daily exercise programme was reinforced. Counselling was provided to address her psychological issues. Orthopedic consultation was given for patellar subluxation and has been suggested further imaging and review. With these interventions, she reported significant pain relief and increased walking ability with an improvement in walking endurance from 5m at admission to about 15m at discharge. She was independent in all her daily activities.

Figures 8: Ambulating with solid ankle foot orthoses and walker

Figures 9: Ambulating with solid ankle foot orthoses and walker Follow-up at 1 year:

She continued to be independent in all her activities and was working as an HR executive with a local company. She reported that her overall quality of life and ambulation had greatly improved with bilateral AFOs and rollator. Fatigue on exertion was much lesser and she reported almost complete relief of her joint pains. She still faced challenges in personal care and hygiene. She started having scapular pain and stiffness over the cervical spine again which was managed with analgesics, muscle relaxants and exercises.

DISCUSSION
There are more than 350 cases of Osteochondrodysplasias / skeletal dysplasias (SD) mentioned in literature. Pseudoachondroplastic spondylo-epiphyseal dysplasia (PSACH) named by Maroteaux and Lamy in 1959 is a type of skeletal dysplasia with a prevalence of 1 in 30,000 individuals [8]. In a PUBMED/ literature review, there were more than 800 studies on SDs, 200 studies differentiating Classic Achondroplasia and PSACH and NO studies on rehabilitation management of PSACH. Table 2 shows the clinical and radiological features differentiating the two entities.

Figures 6: Bilateral ankle instability on weight-bearing
Rehabilitation of persons with skeletal dysplasias may present unique challenges because of their physical stature as well as medical complications. The physical, mental and psycho-social issues along with the stigma that come with dwarfism need to be addressed so that patients living with PSACH as well as other SDs live a normal life. In addition, issues like architectural and attitudinal barriers which such people face in a developing country like India complicate this situation further. Altered lifestyles, modification techniques for ease of ADL, modified surroundings and home environment along with disciplined daily exercise need to be stressed on. Rhizomelic shortening can create disability because of difficulty in reaching the top of the head and the perineum for hygiene care. Disability from shortness of the upper limbs may be exacerbated by flexion contractures of the elbows [9]. Customised orthoses and aids can be prescribed readily if proven to be beneficial. There is sparse evidence and literature regarding rehabilitation of persons with skeletal dysplasias. This limits the ability to provide information, advice and support to those with this condition. More data needs to be collected for effective rehabilitation techniques in different types of SDs.

**CONCLUSION**

Physical, mental and psycho-social challenges can be overcome in a patient with PSACH, Quality of life can be improved: The role rehabilitation medicine plays in this instance is to help patients attain the independence and ability they need to adapt to the difficulties of a life with short stature and other related physical complications. - Precision of diagnosis is important: Pseudoachondroplasia has been mistaken for classical Achondroplasia which is the most common type of short limbed dwarfism. The two, although of genetically different phenotypes, can be mistaken if the differentiation between their presenting features is not clear. The precision of the diagnosis is important as it provides accuracy and meaning to genetic counseling and patient management.

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