HEREDITARY MOTOR AND SENSORY NEUROPATHY (HMSN) DISEASE - IN TWO SIBLINGS
Sunil Kumar Agarwalla* & Nasreen Ali
*Associate Professor, Department of Pediatrics, M.K.C.G Medical College, Berhampur, Ganjam, Odisha-760004, India
Junior Resident, Department of Pediatrics, M.K.C.G Medical College, Berhampur, Ganjam, Odisha-760004, India

Abstract
Hereditary motor sensory neuropathy (HMSN), also known as Charcot-Marie-Tooth Disease, is an inherited, progressive disease of the nerves with weakness and numbness more pronounced in the legs than the arms. Parts of the nerve cells deteriorate. The muscles in the hands and feet get weak because they no longer receive normal impulses from the nerves, and therefore are not being used adequately. Symptoms vary greatly. In addition to muscle weakness, there can be fatigue, pain, numbness, lack of balance, sight and hearing. There can be high arched feet, hammer toes, foot drop, and foot deformities, and possibly scoliosis. This disorder has been scarcely reported in the Indian literature. Here we report 2 such cases from one family who presented to pediatric department of M.K.C.G medical college with complaints of progressive weakness in lower limbs.

Introduction
Slowly progressive distal weakness, muscle atrophy, and sensory loss due to an inherited peripheral neuropathy was described independently in 1886 by Charcot and Marie in France and by Tooth in England[1,2]. A few years later, Dejerine and Sottas recognized and described a more severe, infantile form of inherited neuropathy. [3]Starting in the 1950s, the clinical use of nerve conduction studies combined with pathological information allowed patients to be divided into 2 major groups.[4]
• Group 1 was characterized by slow nerve conduction velocities and evidence of hypertrophic demyelinating neuropathy.
• Group 2 was characterized by relatively normal nerve conduction velocities and axonal degeneration.

In 1975, Dyck expanded the classification system of what was now known as hereditary motor and sensory neuropathy (HMSN) to include forms with additional features. [5]
• HMSN types 1A and 1B (dominantly inherited hypertrophic demyelinating neuropathies)
• HMSN type 2 (dominantly inherited neuronal neuropathies)
• HMSN type 3 (hypertrophic neuropathy of infancy [Dejerine-Sottas])
• HMSN type 4 (hypertrophic neuropathy [Refsum] associated with phytanic acid excess)
• HMSN type 5 (associated with spastic paraplegia)
• HMSN type 6 (with optic atrophy)

Case Report
A 10-year old girl, first seen at 8 years of age, presented to us for evaluation of progressive weakness noticed since late childhood. She was born of a 3rd degree consanguineous marriage and had an uneventful antenatal and perinatal history. There was no family history of similar complaints. There was no complaint of any major illness in the past.
Since she was 5 years of age, her parents noticed that she had distal limb weakness, initially in the lower limbs with frequent slipping of footwear, which progressed leading to gait abnormality and loss of balance. She then gradually developed upper limb distal weakness with difficulty in performing fine motor movements like writing and picking up small objects. There were no complaints of difficulty in rising from a sitting position or in lifting heavy objects. There was no history of sphincteric involvement or of other sensory symptoms. She had a normal intellect and attended school.

On examination, she had lower limb weakness with ankle dorsi-flexion and plantar-flexion power of grade III. Her proximal lower limb power was grade IV at knee flexion and extension, while power at hip flexion and extension was also grade IV. In the upper limb, the distal power at the wrist was grade IV while proximal power in the upper limbs was grade V (Medical Research Council grade). There was areflexia in both lower and upper limbs. Pes cavus deformity was noted at presentation [Figure 1]. Her sensory examination was unremarkable. No other major dysmorphic features were present and the other system examination was normal.

Her younger sibling now of 6 years was starting to have weakness in the lower limb and mild Pes cavus deformity. The power was grade 4 across all the joints of lower limb and deep tendon reflexes were absent.

Their routine hematology and biochemistry including liver function tests, serum electrolytes and blood sugar were normal, creatine phosphokinase (CPK) and lipid profile was normal.

The nerve conduction velocity (NCV) study of the motor nerves in elder sibling showed reduced amplitude without any conduction block or dispersion. The latencies were prolonged with uniform slowing. The conduction velocities were in the range of 50-54 m/S [Figure 2]. Her electromyography (EMG) study showed chronic denervation with partial re-innervation, suggesting a chronic process of demyelination. The NCV of both motor and sensory nerves revealed similar findings in the younger sibling. The nerve conduction screening study in rest of her family was normal.

The parents were counselled about the progressive nature and unavailability of any treatment.
Discussion
HMSN includes includes a clinically and genetically heterogeneous group of disorders which are the most common inherited neuromuscular disorders with an estimated prevalence of one in 2500 individuals[6]. Not only does CMT disease present with a significant genetic heterogeneity but it may also segregate with different Mendelian patterns-autosomal dominant (AD), autosomal recessive (AR) or X-linked.[6] The AR HMSN phenotypes are usually more severe and have an earlier onset of the weakness than the AD HMSN phenotype. [7,8] The clinical and histopathological features in sural nerve biopsies and various genetic studies are needed to identify the exact type of the disease.

Conclusion
Hereditary motor and sensory neuropathies are relatively common and are often inherited with other neuromuscular conditions, and these co morbidities cause an accelerated progression of the disease. But generally they are slowly progressing and the child can have a normal life span but with physical deformities hence physiotherapy is the main stay of treatment.

References
2. Tooth HH. The peroneal type of progressive muscular atrophy. London: Lewis. 1886.