Osteopetrosis is a collective term for a range of sclerosing bone diseases resulting from an absence or defective function of osteoclasts. It was first described in 1804 by Albers Schonberg a German radiologist. The exact incidence is not known, but it is estimated to be 1 in 2,00,000. Inheritance follows either autosomal recessive or dominant patterns, these being diagnosed typically in infancy and adulthood respectively. Intermediate forms are also described which may represent either autosomal recessive disease with milder mutations or early diagnosis of the more severe end of autosomal dominant spectrum.

The clinical expression of osteopetrosis is variable and affected children usually have severe manifestations of abnormal bone remodelling (poor growth, brittle bones, recurrent fractures, delayed tooth eruption, macrocephaly and frontal bossing), deficient and extramedullary hematopoiesis (anemia and hepatosplenomegaly), susceptibility to infections and neurological impairment. The major neurological effects of osteopetrosis result from restriction of growth of foramina, through which the cranial nerves, spinal cord and major blood vessels traverse the skull. Patients may therefore develop optic, facial and trigeminal neuropathies, strabismus, hearing loss, dysarthria, hydrocephalus, cerebral atrophy and developmental delay. The spectrum of neurological involvement is variable depending on the age of onset and severity of the disease. Mildly affected patients may be asymptomatic or develop cranial neuropathies only later in life while children with early onset disease develop progressive cranial nerve problems and hydrocephalus.

Arnold Chiari malformation is a condition characterised by caudal displacement of the cerebellar tonsils through the foramen magnum. It occurs mainly in adults and presents with a wide variety of symptoms and signs suggestive of cerebellar, bulbar or cervical cord dysfunction. The common manifestations include headache, weakness and numbness of limbs associated with hyperreflexia or hyporeflexia, diplopia, dysphasia, dysarthria, nystagmus and ataxic gait.

We report a case of osteopetrosis with Arnold chiari malformation type I with brain stem compression syndrome.

CASE REPORT

A 15-year-old boy presented with headache, giddiness and hoarseness of voice of 6 months duration. The boy was born to a non-consanguineously married couple by full term normal delivery. His developmental milestones were within normal limits and there was no
Osteopetrosis with Arnold Chiari Malformation Type I and Brain Stem Compression

other complaints in the past. There was no family history of anemia, neurological syndrome or early deaths in childhood.

On physical examination, the child was short with dysmorphic features in the form of anti-mongoloid slant of the eyes (Fig. 1). The other features noted were pigeon shaped chest, scoliosis, short hands, short fingers and wide thumb. Neurological examination revealed an I.Q. of 50 (modified Binet Kamath I.Q. Assessment). He had hoarseness of voice, decreased palatal movement and absent gag reflex suggesting involvement of lower motor neuron type of IX and X cranial nerves. There was wasting of tongue with fasciculations suggestive of XII cranial nerve involvement. All the 4 limbs showed spasticity, reduced power (4/5) and exaggerated deep tendon reflexes. There was bilateral patellar clonus and positive Babinski sign. Cerebellar signs like incoordination, positive Romberg’s sign and ataxic gait were noted. Other systems were clinically normal.

vertebral bodies and pelvis (Fig. 2). X-ray of long bones revealed increased bone density with loss of corticomедullary differentiation (Fig. 3). The diagnosis of osteopetrosis was made based on the skeletal survey. MRI Brain showed Arnold Chiari Malformation type 1 with syrinx in the cervical region (Fig. 4).

Fig. 1. Short stature, prominent eyes, antimonogoloid slant of the eyes.

Routine investigations of blood, revealed a haemoglobin of 12 gm%, total leucocyte count of 11,500 / mm³, with 65% neutrophils, 30% lymphocytes and 5% monocytes. Platelet count was 2,05,000 /mm3. Routine urine investigations were within normal limits. X-ray of the skull showed increased bone density and osteosclerosis more so in the base of skull with non pneumatisation of frontal sinuses and sclerosis of mastoid bones. Sclerosis was also noticed in the

Fig. 2. X-ray pelvis showing sclerosis of vertebral bodies and pelvis.

Fig. 3. X-ray of long bones revealed increased bone density with loss of corticomедullary differentiation.