Carpal tunnel syndrome in pediatric mucopolysaccharidoses

Report of four cases

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Four pediatric patients with mucopolysaccharidoses and an associated carpal tunnel syndrome are presented. Findings in these cases were typical of the adult form of median nerve compression at the wrist, but the patients had minimal symptoms in view of these findings. The importance of careful clinical examination and early surgical decompression is emphasized.

KEY WORDS - carpal tunnel syndrome - mucopolysaccharidoses - children

The Hurler syndrome, formerly called "gargoylism" or "lipochondrodystrophy," is a disorder now known to be caused by a defective mucopolysaccharide metabolism. The clinical spectrum of coarse facies, claw hands, dysostosis multiplex, corneal opacities, mental retardation, and death within the first two decades is easily recognized by most physicians. Less well known is the fact that careful clinical, biochemical, and genetic study has revealed a number of different conditions involving abnormal mucopolysaccharide metabolism. Many of these diseases are associated with normal or near normal intelligence and have a better prognosis. Unfortunately, while some of these patients survive longer, they are still prone to many of the irreversible complications of these diseases, for instance, patients must learn to adapt to the claw hand deformity. Loss of thumb function in a hand with already restricted mobility can have significant consequences.

We are reporting the presence of the carpal tunnel syndrome with median nerve compression in four children with mild mucopolysaccharidoses.

Case Reports

Cases 1 and 2

These two brothers were first seen for hand complaints at the age of 10 and 11 years. They had been diagnosed as having mild Hunter's syndrome 6 months before admission. Their mother had noticed decreased joint mobility of the extremities since their early childhood. The boys were very similar in appearance, having rather coarse facies, prominent metopic and sagittal sutures, short necks, systolic ejection murmurs, hepatosplenomegaly, claw hands, and decreased joint mobility at the fingers, wrists, elbows, shoulders, and knees. No corneal opacities were present. They were the shortest members of their school classes, but were competing well academically with their peers.

Neither of the children complained of pain, numbness, or tingling. Examination showed severe thenar atrophy and weakness, and sen-
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Sensory loss to cotton and pin over the median nerve distribution, but Tinel's sign and wrist flexion tests were negative. Median nerve conduction was absent across the wrists. At surgery the carpal ligament, tendon sheaths, epineurium, and periarticular tissues were all thickened. Both median nerves were markedly compressed and cyanotic. One month after surgery, sensation was returning but the marked motor weakness was unchanged.

Cases 3 and 4

These two brothers were first seen in the Genetics Clinic at the ages of 8 and 12 years. They were very similar in appearance, with a broad face and scaphoid skull. Past history revealed that they both had had previous inguinal herniorrhaphies and chronic otitis media. Each had hepatosplenomegaly, peripheral corneal haziness by slit lamp examination, systolic ejection murmurs, and multiple joint contractures including claw hands, limited elbow extension, inability to abduct their arms above their shoulders, and flexion contractures of their toes. Both were in the low range of normal in height. Their bone ages were three to four years behind their chronological ages but they had normal IQ scores. The diagnosis of the Scheie syndrome was confirmed by enzyme assay of skin fibroblast cultures.

Neither child complained of nocturnal pains or tingling. In both boys bilateral thenar atrophy and diminished pin sensation in the median distribution was pronounced. The findings of median nerve compression were more marked in the older boy, and median nerve conduction across the wrist was absent bilaterally in his hands. Median nerve conduction across the wrists in the younger boy was markedly slowed. Incomplete denervation potentials of the abductor pollicis brevis were seen in all four hands.

At surgery the carpal ligaments, tendon sheaths, and periarticular tissues were all thickened. The nerves were all flattened and cyanotic. One month after surgery, all four hands had normal sensation and increased strength. The incisions were all well healed.

Discussion

There are at least 10 different mucopolysaccharidoses. They are all inheritable disorders, characterized by an abnormal accumulation of mucopolysaccharides in connective tissues related to faulty degradation of these substances.

Clinically, these syndromes are relatively similar and all exhibit moderate to severe skeletal and joint deformities, and coarse facies. Mental retardation, dwarfism, corneal opacities, cardiac murmurs, hepatosplenomegaly, inguinal hernias, and skin changes are present in varying degrees. Our patients were characteristic examples of the Scheie and mild Hurler syndromes, as established by urinary excretion studies, by cross-correction experiments with skin fibroblast cultures, and by enzyme assay.

All four patients exhibited classic findings of median nerve compression at the wrist both by physical examination and by electromyographic criteria. However, the usual symptoms of the carpal tunnel syndrome seen in the adult were strikingly absent. None of these boys had any complaints of nocturnal dysesthesias, numbness or tingling, pain on wrist flexion, or positive Tinel's sign. This absence of symptoms clearly points out the importance of careful examination of these children.

Each hand was operated on under general anesthesia through a palmar incision. The eight median nerves all showed varying degrees of flattening, cyanosis, venous engorgement, and thickening of the synovium, the periarticular tissue, and the ligament itself. All incisions healed well, and each patient was improved postoperatively. The results were best in the hands that were initially less affected. Obviously, if optimal function is to be attained, early diagnosis is essential. The failure of these children to complain of the usual symptoms of median nerve compression makes careful surveillance by examination important.

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Reference


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