ATLANTO-AXIAL SUBLUXATION IN THE MORQUIO SYNDROME

Report of a Case

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The Morquio syndrome (mucopolysaccharidosis type IV) is an uncommon genetically
determined disorder of connective tissue. Characteristic features are disproportionate dwarfism
associated with skeletal deformities, particularly genu valgum, pigeon breast and lumbar gibbus. In late childhood paraplegia from cord compression often develops and infiltration
of the cornea and cardiac valves may become evident.

Hypoplasia of the dens (odontoid process) of the axis vertebra is a consistent but less well
recognised feature (Langer and Carey 1966). This abnormality places affected patients at
considerable risk from atlanto-axial subluxation, with consequent damage to the spinal cord.

The purpose of this paper is to describe an affected child who died from tetraplegia of
acute onset following atlanto-axial subluxation during general anaesthesia.

CASE REPORT

The patient, a white girl, was born in 1963 after an uneventful pregnancy and delivery.
Her three elder siblings were normal, as were her parents and other members of the kindred.
There was no known consanguinity.

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In early infancy it was noticed that she had a barrel-shaped chest and later that she was of small stature. During early childhood the skeletal deformities became increasingly evident and she walked with difficulty. However, her mentality remained normal and she suffered neither ocular nor cardiac complications. At the age of six, osteotomies were carried out for bilateral genu valgum. She now had disproportionate dwarfism, with a moderate lumbar gibbus and gross thoracic deformity. Her hands were mis-shapen and loose-jointed, her neck was short, and she had a semi-crouching stance (Figs. 1 and 2).

The dental enamel was defective and generalised hirsutism was present. The nose was stubby, the mouth broad and the teeth widely spaced; nevertheless, her features were not ugly. The corneae were clear. There were no cardiac murmurs and the liver and spleen were not enlarged.

Radiology of the spine revealed vertebral bodies which were flattened, with anterior projections and wide disc spaces (Fig. 3). The pelvis was long and narrow, with flaring of the iliac wings. The sternum protruded and the lower ribs were broadened anteriorly. The upper and lower femoral epiphysial centres were fragmented and compressed and there was moderate genu valgum. At the distal end of each femur the growth plate was sclerosed and trabeculations were increased in the femoral shaft.

Examination of the urine revealed an excess of mucopolysaccharides, and on a basis of this finding, together with the characteristic clinical and radiological features, a diagnosis of mucopolysaccharidosis type IV was made.

Progress and treatment—Walking became increasingly difficult and in August 1971 signs of upper motor neurone involvement were evident in the lower limbs. It was considered that spinal cord compression might be responsible and myelography was undertaken under intravenous anaesthesia. She remained unconscious for several hours and on awakening was found to be tetraplegic, all sensibility and movement being absent below the level of the neck. Tracheostomy and intubation were performed and mechanically assisted respiration was initiated. Radiographs revealed that the atlas vertebra was now displaced forwards on the axis and it was concluded that damage to the spinal cord had occurred at this level, from excessive movements of the head during anaesthesia (Fig. 5).

The dislocation was reduced by neck traction, but the upper cervical spine remained unstable and neither sensibility nor muscle power returned. Although artificial ventilation was continued, she developed frequent periods of anoxia which were accompanied by abnormalities of cardiac rhythm. Her condition deteriorated and she died fourteen days after the initial event.

At necropsy pulmonary infection was found to be the immediate cause of death. The aortic and mitral valves were found to be irregular and heavily infiltrated with mucopolysaccharide material. The liver, spleen and kidneys were microscopically normal. As permission...
had been obtained for only a limited examination, neither the cervical cord nor the brain was accessible for study.

**FIG. 4**

Figure 4—Lateral radiograph of the cervical spine ten weeks before the onset of tetraplegia. Although the cervical vertebrae were flattened and mis-shapen, the atlas and axis vertebrae bore a normal relationship to each other.

**FIG. 5**

Figure 5—Lateral radiograph of the cervical spine after the onset of tetraplegia. Note the forward shift of the atlas on the axis.

**DISCUSSION**

Although the eponym “Morquio syndrome” is frequently applied to any dwarf with spinal abnormalities, the term is more properly used to denote the patient with mucopolysaccharidosis type IV who has distinctive clinical features and excess excretion of keratosulphate in the urine.
(McKusick 1966). This distinction is important because precise diagnosis permits accurate prognosis and rational management.

Genu valgum and paraplegia of slow onset due to spinal cord compression are common complications which may bring children with the true Morquio syndrome to the orthopaedic surgeon (Einhorn, Moore and Rowntree 1946; Hobaek 1961). Affected patients develop valvular and myocardial infiltration during late childhood, and usually die in early adult life (McKusick, Kaplan, Wise, Hanley, Suddarth, Sevick and Maumenee 1965). For this reason, and in view of the inherent risks of operation, major surgery may not be considered to be justifiable. Nevertheless every patient must be considered in the light of his or her own particular situation.

When an operation is to be undertaken, it is important that hypoplasia of the dens and instability of the cervical spine are taken into account. If the patient's predisposition to atlanto-axial dislocation is recognised, excessive head movements during general anaesthesia can be avoided. In this way, disastrous complications can be prevented.

**SUMMARY**

1. The case is described of an eight-year-old girl with the Morquio syndrome (mucopolysaccharidosis type IV) who died from acute tetraplegia, due to atlanto-axial subluxation which occurred during general anaesthesia.

2. Hypoplasia of the dens of the axis vertebra entails a high risk of vertebral dislocation and spinal cord damage especially in circumstances such as general anaesthesia when excessive movements of the head may occur.

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**REFERENCES**


