Morquio Syndrome: A Rare Case Report

Vidya Sagar Mopagar, Mithesh D Kathariya, Umapathy T, Premkishore K, Chandrashekar, Ashish Raurale

ABSTRACT

Morquio’s syndrome is an autosomal recessive disorder caused by deficiency of N-acetylgalactosamine-6-sulfate. It was first described in 1929 by Morquio and Brailsford. Morquio’s syndrome is characterized by a defect in the degradation of keratin sulfate resulting in the accumulation of mucopolysaccharides. At birth, a patient with Morquio’s syndrome may appear healthy, however as the child grows into adulthood, various manifestations of this syndrome begin to emerge. Diagnosis is typically based on clinical examination, radiographs, urinary GAG tests, and the enzymatic activity of N-acetylgalactosamine-6 sulfatase in blood cells or fibroblasts. Once it is diagnosed, MS requires a multidisciplinary approach to patient care.

Keywords: Morquios syndrome, Autosomal disorder, Multidisciplinary approach.


Source of support: Nil

Conflict of interest: None declared

INTRODUCTION

Morquio’s syndrome also known as mucopolysaccharidosis type IVA is an autosomal recessive disorder caused by deficiency of N-acetylgalactosamine-6-sulfate. It was first described in 1929 by Morquio and Brailsford. The incidence is unknown but is estimated to be between 1 in 75000 population in Northern Ireland to 1 in 200,000 population in British Columbia 1 in 640,000 live births in Western Australia. Morquio’s syndrome is characterized by a defect in the degradation of keratin sulfate resulting in the accumulation of mucopolysaccharides. At birth, a patient with Morquio’s syndrome may appear healthy, however as the child grows into adulthood, various manifestations of this syndrome begin to emerge, including coarse facial features, prognathism, a broad mouth, and a short nose with anteverted nares and a flat nasal bridge, widely spaced teeth and macrocephaly. Other features include aortic valve incompetence, hepatomegaly, inguinal hernias, mixed hearing loss and ocular complications including clouding of the corneas, pigmented degenerative retinal lesions or glaucoma. Pulmonary complications include a restrictive defect due to kyphoscoliosis resulting in decreased lung volumes, ventilation-perfusion mismatching and obstructive sleep apnea, which can result in pulmonary hypertension and cor pulmonale. Characteristic vertebral abnormalities include anterior hypoplasia of T12, L1 or L2, which may give rise to lumbar kyphosis. Hypoplasia of the dens is a common and severe manifestation that may lead to atlantoaxial instability, compression of the cervical spinal cord, and complications during endotracheal intubation. The limb-bone abnormalities may include short diaphyses, curving of the metaphyses and poor development of the epiphyses. Pelvic abnormalities include widening of the acetabula, hypoplasia of the femoral heads, with valgus deformity of the femoral necks. Diagnosis is typically based on clinical examination, radiographs, urinary GAG tests, and the enzymatic activity of N-acetylgalactosamine-6 sulfatase (GALNS) in blood cells or fibroblasts. Once diagnosed, MPS IVA requires a multidisciplinary approach to patient care.

CASE REPORT

A 15-year-old boy reported to the Department of Pedodontics, Rural Dental College, PMT Loni with a chief complaint of decayed teeth. Starting from the entry in to the dental office the child gained everyone’s attention by his gait and behavior. The child had a waddling gait and was bent on one side. He was speaking loudly, interestingly and was quite co-operative but at times adamant in communication. History did not reveal any familial incidences of similar disorders. The child was normal in milestones till he was 7 years old and later the symptoms of abnormal behavior started becoming apparent. General body examination revealed long face with a short neck with jerky movements. The chest showed typical pigeon chest deformity with marked increase in anteroposterior dimension accompanied by abdominal distension (Fig. 1). The external genitalia showed descended testis. Extraorally the nose was long and broad with nares placed quite apart. Ears were placed slightly inferiorly and were large in size. The midface was very prominent with broad mouth (Fig. 2). Intraoral examination showed multiple decayed teeth with over retained root pieces of primary teeth. The maxillary left lateral incisor and both the first molars in the mandibular arch were cariously destructed (Fig. 3). The patient reported frequent painful episodes with the destructed teeth and had difficulty in eating. An OPG was advised to assess the status of development of the teeth.
and treatment planning which revealed, slightly delayed
dental development as the roots of many teeth were still
with open apices.

Extraction of the root pieces was planned. Since the
lower left first molar was nonrestorable due to carious
destruction it had to be considered for extraction.
Endodontic treatment with upper left lateral incisor and
lower right molar followed by crowns was planned. The
patient was referred to pediatrics for examination and
consent for treatment. It was then decided to perform entire
procedures under general anesthesia and accordingly the
treatments were performed.

Although, the dental part of management was
uncomplicated, inducing general anesthesia was difficult.
It was noticed that the size of the epiglottis was abnormally
large and the trachea was small making the endotracheal
intubation very difficult. The anesthetic team had to be
extra alert to manage any complication if exist. The
anesthetic recovery was disturbing and traumatic with mild
bleeding which later stopped. The child recovered
completely from anesthesia but was slightly irritated
warranting additional observation.

**DISCUSSION**

Mucopolysaccharidosis (MPS) are a family of inherited
metabolic diseases that results from the deficiency of
lysosomal enzymes involved in the degradation of the
glycosaminoglycans (GAGs; mucopolysaccharides). These
GAGs include dermatan sulfate (DS), heparan sulfate (HS),
and keratan sulfate (KS). Chondroitin sulfate (CS) may also
be involved. On the basis of clinical and biochemical studies,
these disorders have been designated as MPS I through
MPS VII.⁶

Among many syndromic patients Morquio’s syndrome’s
presentation is slightly complex. The condition is not very
commonly seen and difficult to diagnose in the initial few
years of life. Most of the children with Morquio’s syndrome
are normal in their appearance during first few years of life
making the condition less noticeable. A familial tendency
of unknown etiology has also been reported by Rekha et al
(2012) where three siblings in the same family were affected
with the syndrome.⁷ Certain dental abnormalities specific
of Morquio’s syndrome among all types of mucopolysac-
charidosis include presence of thin enamel with dentin
being visible, pitting and hypoplastic defects, Sharp pointed cusps and weak enamel. However, no prominent dental abnormalities were evident in the present case.8,10 Present case did not have any complicated dental treatments apart from routine procedures, but the danger faced was during inducing and reversal of general anesthesia. Anesthetic implications of Morquio syndrome similarly in majority of the cases might relate to end-organ dysfunction and anatomical distortions related to the intracellular accumulation of keratin sulfate.11 Difficulty in intubating the trachea could have been as a result of a number of features in particular, atlantoaxial instability and hypoplasia of the odontoid process, etc. Additionally, bulky pharyngeal soft tissue due to the deposition of mucopolysaccharides in the soft tissues of the oropharynx, floor of the mouth, epiglottis, aryepiglottic folds and macroglossia may mandate the use of a smaller endotracheal tube, furthermore the presence of prominent maxillae, limited mouth opening due to involvement of the temporomandibular joints and a short neck makes safe direct laryngoscopy difficult to perform. All of these factors may lead to a ‘cannot intubate/cannot ventilate’ scenario which is the greatest challenge even for the anesthetists. Furthermore, cervical spine instability in these patients is often not confirmed or excluded by adequate radiographic examination and functional clinical testing and cervical spine stability may not be preserved in the deeply anesthetized patient with neuromuscular blockade.12 Hence, keeping in mind the possibility of unexpected complications a pediatric anesthetist must be well trained and skilled for such situations and a multidisciplinary management approach is warranted. Enzyme replacement of GALNS (called BMN 110), which is designed to clear keratan sulfate from the lysosome has also been implicated to slow down the disease progress and reduce the signs and symptoms.

REFERENCES


ABOUT THE AUTHORS

Vidyasagar Mopagar (Corresponding Author)
Professor, Department of Pedodontics, Rural Dental College, Pravara Institute of Medical Sciences, Loni, Maharashtra, India, e-mail: vidyasagar@gmail.com

Mithesh D Kathariya
Senior Lecturer and PhD Student, Department of Pedodontics Rural Dental College, Pravara Institute of Medical Sciences, Loni Maharashtra, India

Umapathy T
Reader, Department of Pedodontics, Krishnadevaraya College of Dental Sciences, Bengaluru, Karnataka, India

Premkishore K
Reader, Department of Pedodontics, New Horizon Dental College and Research Institute, Bilaspur, Chhattisgarh, India

Chandrashekar
Senior Lecturer, Department of Pedodontics, Rural Dental College Pravara Institute of Medical Sciences, Loni, Maharashtra, India

Ashish Raurale
Postgraduate Student, Department of Pedodontics, Rural Dental College Pravara Institute of Medical Sciences, Loni, Maharashtra, India