CASE REPORT

The approach to the case of rarely seen adult morque syndrome at emergency service

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ABSTRACT

Acute abdomen examination, its diagnosis and treatment include some difficulties at emergency clinics. The group of patients, who have self-expression problem, is also one of these difficulties. Because of the opportunities available in today's medicine, patients who have severe congenital malformations have increased survival, so, such kinds of patients can also apply to adult emergency clinics. Diagnosis, follow-up and treatment phases of this patient group present quite a lot of differences when compared to other patient groups. The aim of this presentation was to discuss a case of morque syndrome patient who came to our emergency clinic with the complaints of nausea, vomit, nuisance for 4 days and got the diagnosis of retangule umbilical hernia with literature.

Key words: mucopolysaccharidosis IV, umbilical hernia, emergencies.
INTRODUCTION

Morquio syndrome (MPS IV), which is a kind of mucopolysaccharidosises, is a hereditary lysosomal storage disease. Such patients generally pass away due to cardiopulmonary and neurologic function disorders at the age of young adults (1). However, medical care and close clinical observation developing recently have extended the life time of the patients until the sixth decade (2). Even though it is so rare, adults who have congenital genetic disease apply to adult emergency clinics as a result of these developments. It is aimed in this case to present a case of morque syndrome patient who came to our emergency clinic with the complaint of abdominal distension and got the diagnosis of etrangule umbilical hernia.

CASE REPORT

The male patient who was 27 years old and had morquio syndrome (MPS IV) was brought to our emergency clinic by his family due to the complaints of nausea, vomit, abdominal distension and inability to defecate for 4 days. It was learnt that the patient who had dysmorphic appearance (Figure 1) had been diagnosed when he had been 4 years old and was followed due to deficiency in motor development, critical hearing loss, heart failure and chronic bronchitis. The communication with the patient was enabled by the help of his family due to hearing loss and dysphasia.

While vital findings were stable, tension arterial was 100/60 mmHg, pulse was 85/minutes, axillar fever was 36.7°C, respiratory rate 20/minutes. Abdomen distension and umbilical herniation were diagnosed at systemic physics examination (Figure 1). Skin texture of umbilical sac had an ecchymotic appearance and there were common air-fluid levels at direct abdomen graphic ambulant. In the genetic examination, advanced short stature, pectus carinatum, coxa valga and bilateral genu valgum deformities, closing in the proximal of metacarpal bones, thickening in distal, distal humerus and abnormal aspect in ulna were observed related to dolichocephaly, spondyloepiphysial dysplasia. No pathology, except for bowel anses drowned in umbilical herniation, was encountered in abdominal ultrasonography made with the aim of externalizing organic anatomic organ anomaly. As a result of general surgery consultation, etrangule umbilical hernia was detected and operation decision was taken. Necessary preparations for difficult intubation were made due to existing dysmorphologic appearance, mallampati score 4, restriction of mouth opening and big tongue. Firstly, the operation of the patient was performed with successful spinal anesthesia instead of planned regional anesthesia. The patient who did not need postoperative intensive care was sent to general surgery service without any problem.

Mucopolysaccharidosis (MPS) is a disease group composed of deposit of partly broken mucopolysaccharids at lysosomal storages. These residues accumulate in lysosomes as a result of deficiency of one of enzymes which...
demolish sulphate and carbohydrate residues known as specific hydrolases. Some failures of heparan sulphate, dermatan sulphate, keratan sulphate or chondroitin sulphate may go wrong. Specific enzyme assay is important for detecting MPS type (3,4).

Morquio Syndrome, which is a type of MPSs, was defined by Morquio and Brailsford in 1929 for the first time (5,6). Incidence of Morquio Syndrome is 1/40,000 and it is an autosomal recessive transitive disease. It is characterized with chondroitin-6-sulphate and keratan sulphate storage. It has 2 subgroups characterized with type A, N-acetylgalactosamine-6 sulphate and type B, beta-galaxydos deficiency (7). It is a rarely seen MPS which is shown with the gastrointestinal indications, heart and lung diseases, hearing problems and especially skeletal dysplasias (8). There were skeletal disorders, hearing and cardiac respiratory system problems in our case. The diagnosis is generally made when the patient is 3-4 years old. While the patients who have the subtle form of this disease live until the age of 30-40, those with severe disease die at early adolescent phase (2).

It was learnt in our case that Morquio Syndrome Type A diagnosis was made when the patient was 4 years old. Umbilical and inguinal hernias are frequently encountered in the cases of Morquio Syndrome (9). Intense observation is suggested. Montano et al. reported in their study that approximately 6% patients with Morquio Syndrome Type A were operated when they were averagely 8 years old due to herniation (1). In our case as in the literature (1,9) umbilical herniation is detected as congenital, however, early surgery is not planned but followed up because of the existing dysmorphic appearance, short neck and mouth nature.

Acute nuisance is one of the most frequent reasons among adult emergency service applications. Being able to express the problem of the patient is quite important for the diagnosis by a clinician (10). However, both hospital applications are late and serious difficulties occur at diagnosis phase when the patient is not able to express the problem (10). The big part of this patient group consists of the patients with congenital genetic malformation. It is reported in the study of Khalid ve Al-Salamah that the most frequent emergency application reasons are nausea, vomit, hoove among the patient group who are not able to express their problem due to genetic diseases and the length of hospital application with these complaints is $2.7 \pm 1.0$ days (approximately, 1-5 days). Besides, their postoperative mortalities were found quite high as 33.3 % (10). It was detected in our case as in this study that nausea, vomit and hoove were at the forefront and the patient was brought to our hospital at the end of the fourth day due to lack of expression.

In conclusion, development of early diagnosis methods, early start of treatment, progress of rehabilitation studies improve the patients' quality of life who have severe genetic diseases. As a result of these developments, the patients can apply to the clinics, especially to adult emergency clinics while these cases were just followed up by kid clinics in the past. One of these clinics are adult emergency clinics. However, in today's world, there is no algorithm or enough data to follow up for the approach to this kind of the patient group. We believe that this issue has to be stressed and approach to the patients by creating algorithm would be more appropriate.

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REFERENCES


