

Letter to Editor / Editöre Mektup

**MANAGEMENT OF A PATIENT WITH XERODERMA PIGMENTOSUM FOR
ESOPHAGEAL DILATATION**

Xeroderma Pigmentosum Olan Bir Hastada Özofagus Dilatasyonu Yönetimi

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Dear Editor,

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Fourteen years old female patient was admitted to pediatric surgery clinics for swallowing problems. She had been diagnosed as xeroderma pigmentosum in another hospital and had been treated for dermatological problems. She had mental retardation and orthopedic problems like scoliosis. She could not walk by herself and wheelchair was used for her mobilization. No treatment for neurological problems had been given. Swallowing difficulties began nearly 2 years ago. First she could not swallow solid meals. The problem progressed and when she was admitted to our hospital she could only swallowed liquids for 2 months. She was hospitalized and treated with intravenous nutrition support solutions at first. Water soluble contrast meal for pharynx and upper esophagus graphics was performed and upper esophageal sphincter dystonia was diagnosed. Esophageal dilatation with general anesthesia was suggested to be the best way to choice in the patient's circumstance. Anesthesia induction with thiopental, fentanyl and vecuronium were administrated intravenously. For maintenance, O₂/N₂O mixture and sevoflurane according to patient weight and age was used. Esophagoscopy was found to be normal for anatomic abnormalities and dilatation was performed. Patient waked up without problem and discharged from the hospital with well swallowing of soft meals. She could not take solids because of severe dental problems. In 6 months of follow-up, parents has not observed worsening in her swallowing.

Xeroderma pigmentosum is a rare disease and inherited as an autosomal recessive trait (1). In Xeroderma pigmentosum, there are genes called A to G and some of these genes involved only in nucleotide excision repair (XPA and XPC) whereas the others are not only in nucleotide excision repair but also implicated in other processes including transcription and recombination (1). Xeroderma pigmentosum phenotype can result from the defect in a part of these seven classic nucleotide excision repair (1). In Xeroderma pigmentosum, as there is an acute photosensitivity characterized by sunlight induced abnormal pigmentation, skin is usually like a dry parchment and precocious cutaneous lesions are seen (1). Besides, about 30% of Xeroderma pigmentosum patients have progressive neurological

degeneration (2). Neurological findings for Xeroderma pigmentosum contain peripheral neuropathy, sensorineural deafness, microcephaly, cerebral dysfunction, ventricular dilation, cortical atrophy, basal ganglia and cerebellar disturbances (3). Swallowing difficulties may be important and gastrostomy may be necessary in late stages (2). Developing including abnormal gait, and difficulty walking leading to the necessity of a wheelchair or to quadriplegia are other problems (2). *Muto et al* reported laryngeal dystonia in their case with marked inspiratory stridor (4).

There are controversies with the anesthesia for these patients. *Masuda et al.* managed their Xeroderma pigmentosum patient with total intravenous anesthesia technique as they observed DNA strand breaks in lymphocytes of two Xeroderma pigmentosum patients with halothane exposure *in vitro*(5). *Miyazaki et al* accepted the same point of view and managed their patient with this warning and used intravenous anesthesia (6). As *Brunner et al* reported the evidence is not too strong to avoid all inhalational anesthetics (7). We preferred sevoflurane anesthesia with fentanyl and vecuronium as Brunner and be careful for the contraindications of these drugs.

Although gastrostomy was suggested to be a choice for swallowing difficulties of these patients, we could not find a report giving the criterion of the problem. Besides, up to now, xeroderma pigmentosum and esophageal dystonia has not been reported yet. We experienced that patients in this situation could benefit from esophageal dilatation. Also, there has not been much reports giving information in detail for anesthesia management but we suggest that management of these patients within standard anesthetic procedures are not seemed to be harmful in early periods.

Best Regards,

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