# Fucosidosis and anesthesia

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## ABSTRACT

Fucosidosis is a rare, autosomal recessive lysosomal storage disorder caused by a severe deficiency of α-L-fucosidase. Patients usually have some problems with glycoprotein storage in the brain and other organs, and some structural abnormalities that need special consideration in anesthesia. It has 2 types, the early onset or infantile, and the juvenile. Here we present an 8-year old girl with deformities in the maxillofacial region, with big tongue, small and retracted chin, saddle nose, and short neck that could not be extended, causing difficult intubation, and congenital cardiac problems requiring a special anesthetic strategy.

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Fucosidosis is a rare autosomal recessive lysosomal disorder, secondary to severe deficiency of α-L-fucosidase. It has 2 types, the infantile (60%), with rapid course, clinical signs within the first year, death by 5 years of age; it appears with psychomotor retardation, weakness, hypotonia, spastic quadriplegia, and mental retardation. The other is the juvenile type (40%), which has a late onset at age 2, with tortus conjunctiva vessels, slight corneal clouding, pigmented retinopathy, dark purple telangectasia, and hyperhydrosis.[1] The disorder is associated, in both types, with congenital heart defects, short status, hepatomegaly, macroglosia, coarse skin, and spasticity. The disease can be accompanied by metabolic and gonadal disorders such as hypothyroidism.[1,2] The α-L-fucosidase (EC 3.2.1.51), are widespread glycosidases involved in many biological processes such as inflammation, metastasis and the lysosomal storage disease fucosidosis.[3] Partially degraded glycosaminoglycans are stored in lysosomes and excreted in the urine, and the patients' urine are positive for fucosidase derivatives.[4] Bone marrow transplantation as an effective treatment of central nervous system disease in globoid cell leukodystrophy, metachromatic leukodystrophy, adrenoleukodystrophy, mannosidosis, fucosidosis, and other glycoproteinoglycane storage disorders has been previously proposed.[5] Anesthetic management in patients with fucosidosis needs special consideration, which has not been discussed in previous researches. As a result, the purpose of this study was to describe the anesthetic management in an 8-year-old girl with fucosidosis.

**Case Report.** An 8-year old girl presented for preoperative evaluation for glaucoma surgery. Her current medical history was also remarkable for recurrent pulmonary infections. She had a history of 3 previous operations: corneal transplantation due to keratoconus 5 years ago, tonsillectomy 3 years ago, and hiatal hernia repair, twice the previous year. Her family history revealed that she was the second living child from a consanguineous marriage; she had an older, normal sister. Her IQ score was average, and she had a successful year of study. She had developmental delay (80 cm height and 14 kg weight), and was proposed a short status child. In addition, she had congenital heart defects. On physical examination, she had coarse skin especially on the face, had a small and retracted chin, and a short neck with extension limitation. She had exophthalmic eyes, both were hypermetrope with 1/10 right, and 2/10 left visions. She had a saddle nose and right side choanal atresia, normal oral cavity and palatal arch, with temporomandibular joint movement limitation preventing her from opening mouth properly, with open mouth diameter of 1.5 cm, Mallampathy Grade 4, and 3 cm thyromental diameter, making intubation difficult. She had some joint deformities on her fingers, knee, and arm, with motion range

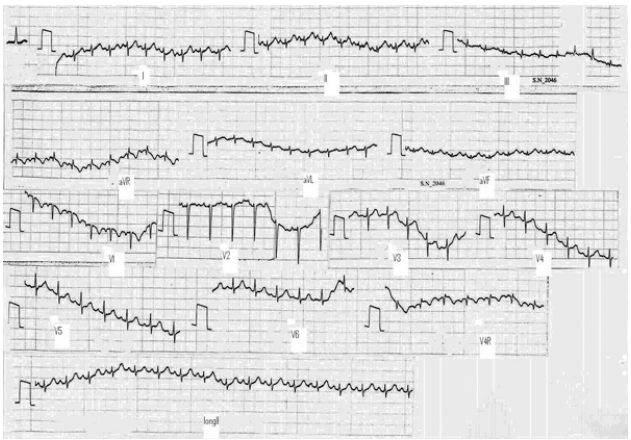

**Figure 1 -** Electrocardiogram showing slight left axis deviation, normal sinus rhythm, and no ST-T change, or arrhythmia.

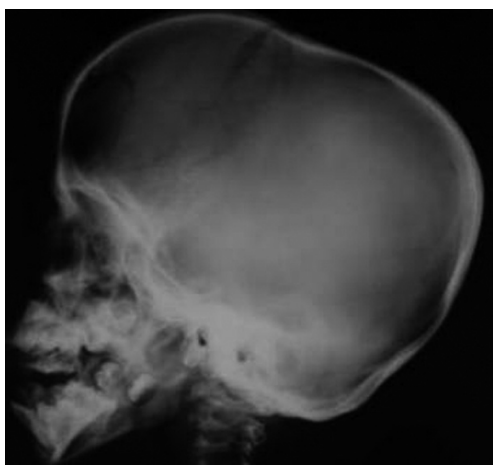

**Figure 2 -** The skull x-ray showing brachycephalia.

limitation. Her blood pressure was 140/60 mm Hg, and she had a systolic and diastolic murmur that suggested mitral regurgitation (MR), aortic insufficiency (AI), mitral stenosis (MS), and aortic stenosis (AS). On the abdomen, the liver span was 12 cm (4 cm under rib edge), with a palpable spleen. Her echocardiography showed thickening of mitral and aortic valves, mild MR and AI, good left ventricular function, with left sided aortic arch. Her electrocardiogram showed a slight left axis deviation, normal sinus rhythm, and no ST-T change, or arrhythmia (**Figure 1**). The radiologic findings revealed protruded forehead (brachycephalia), small and retracted chin, short neck, and a small odontoid process, with atlantoaxial subluxation. A big sella torsica with J-shape skull was shown on x-ray

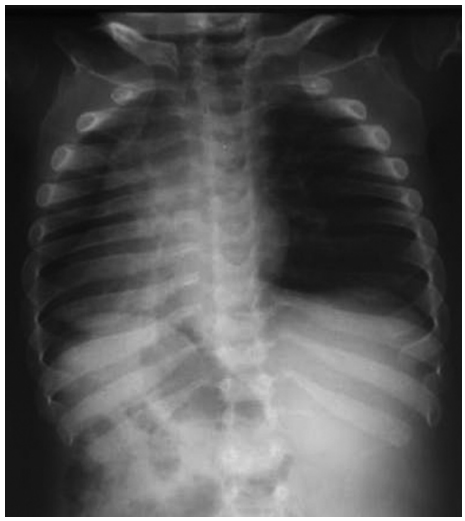

**Figure 3 -** The chest x-ray showing mucopolysaccharidosis.

(**Figure 2**). The chest x-ray showed oal-shaped ribs, dysphasic scapula, and the clavicles were hypoplastic on the lateral head. Epiphyseal deformity is obvious, and is similar to osteal congenital deformities that can be seen in mucopolysaccharidosis (MPS) (**Figure 3**). Laboratory findings showed sufficient gangliosidase and deficient fucosidase in blood. The MPS was negative in urine, and the blood ammonia level was 70.5 mmol/Lit (normal range: 10-47 mmol/Lit).

***Anesthetic management.*** All needed equipment were prepared for this difficult intubation. *Premedication:* One µg/Kg fentanil, 0.1 mg/kg dexamethasone, and 1 mg/kg lidocaine were used in order to reduce the intubation stress, and inflammation of post operation. She was well ventilated with mask during this time. *Induction:* A 5 mg/kg sodium thiopental, and 1.5 mg/kg succinylcholine were used for muscle relaxation. Laryngoscopy was difficult because she had a short neck with extension limitation, and the mouth could not be openned normally. Epiglottis was not apparent on the third try, and it was wide and thick like an adults'. She was intubated with 5.5 mm diameter endotheracheal tube, using stylet in the last try. The lung secretions were suctioned after intubation. *Maintenance of anesthesia:* A low dose halothane with fentanil was used for maintenance of anesthesia, and she was on spontaneous breathing. She was extubated after complete awareness, and oropharyngeal and lung secretions were suctioned; she had a mild vocal cord spasm at extubation that was treated with positive pressure mask ventilation. The recovery was uneventful, and she was sent to the ward in a very good, and stable condition.

**Discussion.** Fucosidosis is an autosomal recessive lysosomal storage disease, characterized by nearly complete deficiency of α-L-fucosidase.[6] The deficiency of the very gene results in accumulation of a variety of fucose-rich glycoproteins, glycolipids, and mucopolysaccharides within the lysosomes of cells of most organs, including liver, spleen, kidney, heart, brain, peripheral nerves, and skin.[7] The reported case fits the picture of fucosidosis. To our knowledge, this is the first paper reporting the anesthetic management in such patients. Most of the lysosomal enzymes involved in the hydrolysis of glycoprotein carbohydrate chains are exol-glycosidases, which removes terminal monosaccharide. Thus, the deficiency of a single enzyme causes the blockage of the entire pathway,[8] and causes problems in hepatic function. Therefore, the use of high dosage of inhaled anesthetics such as halothane, isoflurane, and enflurane, and high dose opioids are not recommended in these types of patients. [9] It could be concluded that in fucosidosis, like other glycoprotein storage disorders, the presence of facial and mandibular deformities makes intubation difficult. These patients also need special anesthetic consideration, because of cardiovascular and liver abnormalities. It should be noted that further studies are required in order to assess different aspects of the anesthetic management in such patients and also to achieve an acceptable protocol.

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