Objectives: Mucopolysaccharidosis (MPS) is a rare, hereditary lysosomal storage disease, which develops due to a deficiency in enzymes that play a role in the metabolism of primary glycosaminoglycans (GAG). Otorhinolaryngological problems in MPS patients may include tonsil and adenoid hypertrophy, as well as tongue hypertrophy causing upper airway obstruction, otitis media with effusion, chronic otitis media, hearing and balance problems and voice disorders. Patients with MPS have serious complications, high rate of morbidity and mortality during the otorhinolaryngological operations.

Methods: We evaluated 50 patients whom we have been following in Hacettepe University Medical Center with MPS disease of all types, with their otorhynolaryngologic, endoscopic and audiometric examinations. We also evaluated computerized dynamic posturography findings of 15, acoustic voice analyze findings of 10 patients with MPS disease.

Results: 50% of the patients had abnormal tympanometric findings. 15% of the patients had sensorineural hearing loss (SNHL), 35% with conductive type of hearing loss (CHL), 20% had mixed type hearing loss and 30% had normal hearing during evaluation of pure tone audiogram. 16% of them had ventilation tube placement. 6% had perforated ear drums. Vestibular dysfunction was also demonstrated in 10 of 20 (50%) of the cases whom vestibular tests could be run. 12 of 18 tested patients (67%) had abnormal acoustic parameters. Airway obstruction and sleep disorder were also present in 60% of them and 22% were undergone tonsillectomy, 40% adenoidectomy and 2% underwent tongue reduction operations. We had lost one patients early post-op period after T&A surgery and in additional 2 patients we had to perform tracheotomy after T&A surgery. Complications increase in time as the disease progress and cause morphologic changes in airway in untreated patients.

Conclusions: ENT surgeons are important for MPS patients. Early diagnosis is very important for them to have a much better quality of life, and we can diagnose MPS very early, as we see them, very early in their life span with frequent ENT infections. 1) Keep in your minds that, early frequent attacks of ENT infections (or ENT surgery history) plus presence of hernia (or hernia surgery history) in a young kid may be due to MPS, and refer these kids to a metabolic specialist. 2) Follow MPS patient very close and try to operate them as early as possible before morphologic changes due to disease becomes prominent and complication rates inevitably become so high, because airway, pulmonary and skeletal functions seriously deteriorates. 3) Multidisciplinary team approach and “being prepared” is vital to prevent complications in MPS patients.