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MINIA UNIVERSITY
FACULTY OF MEDICINE
E.N.T. DEPARTMENT

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**EAR, NOSE AND THROAT DISORDERS
IN CHILDREN WITH DOWN
SYNDROME**

ESSAY SUBMITTED FOR PARTIAL FULFILLMENT OF MASTER
DEGREE IN EAR, NOSE AND THROAT

٣١١٥٥٥

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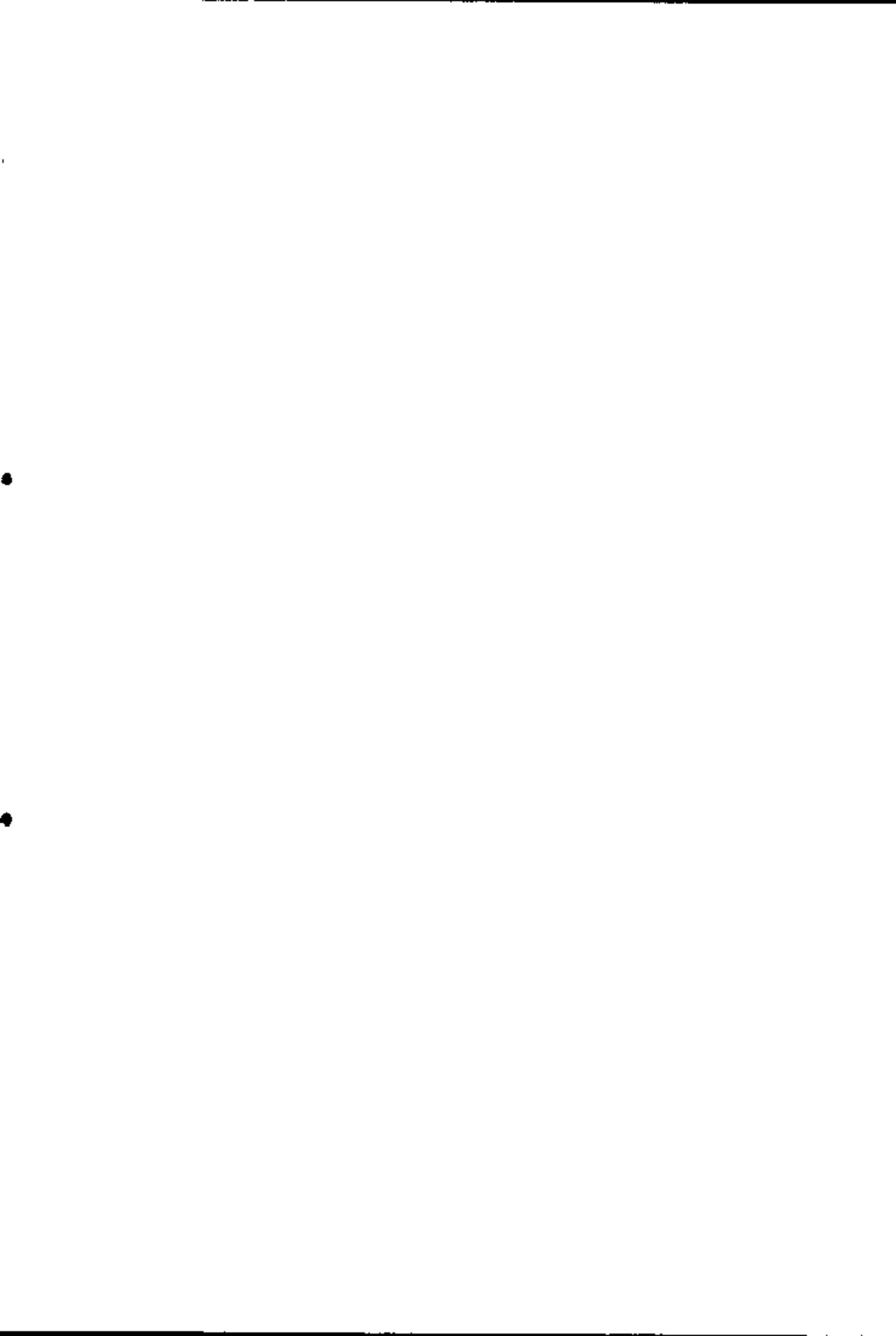
Finally, I hope that this work will be helpful for my colleagues.

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Introduction

Down syndrome is the most autosomal aberration syndrome that was described by John Langdon Down in 1866. (*Nyhan, 1983*) occurring in approximately 1 in 600 live births in England and Wales. (*Bain and Ellis, 1984*).

The incidence however increases significantly with advancing maternal age. In women under 30, the incidence is about 0.6 in 1000 births; this increase to approximately 20 in 1000 births for mothers aged 45 and over. (*Evans and Glass, 1976*)

It is the one of the commonest congenital causes of severe mental retardation. (*Elliot et al, 1981*) In fact, Down syndrome is the primary cause of retardation when the diagnosis is made at birth.

Down syndrome is the result of a trisomy of chromosome 21 or the translocation of the q22 segment to another chromosome. This has led some investigators to conclude that the q22 portion is responsible for this disease. Our incomplete understanding of the genetic abnormality allows for only limited patho-physiologic correlation between the chromosomal aberration and the disease process. Two enzymes involved in purine metabolism, GARS and AIRS, have been mapped to chromosome 21. As altered purine metabolism is related to mental retardation, this could explain the lowered intellectual capacity of these patients. Superoxide dismutase (SOD), known to play a role in aging and carcinogenesis, is also found on chromosome 21 and is a potential etiology for the diminished life span and the higher incidence of leukemia in this population. (*Strome and Strome, 1992*)

The clinical manifestation of Down syndrome are numerous but none of the features other than the mongloid facies are constantly present. Generally mental retardation and hypotonia characterize Down syndrome, which is present since birth. The craniofacial region is characterized by brachycephaly or flat occiput. There is an upward and outward slant of the eyes, which have prominent internal epicanthal folds, speckled irides (Brushfield spots). The nasal bridge is flattened and there may be a high arched palate, large tongue and malformed ears. (Figure 1) At birth there is often an absence of the breast tissue that is normally present in infants secondary to maternal hormonal stimulation. The hands are often short and there is frequently a short incurved fifth finger secondary to the absence or dysplasia of the middle phalanx. There is often single palmar crease known as simian line and a distally displaced triradius. Spacing between the first and second toes with a deep planter

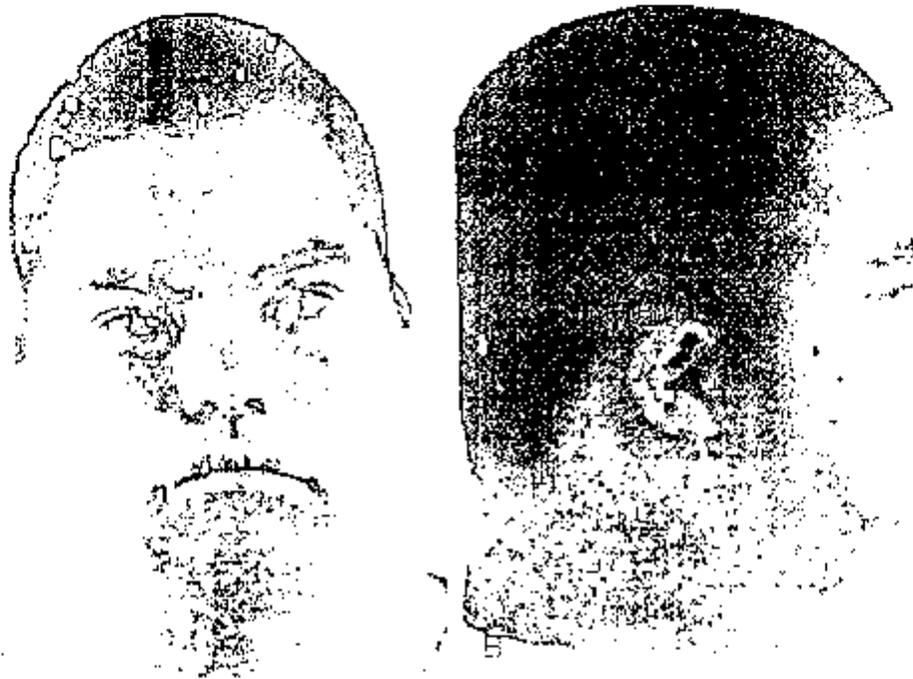


Figure 1. A. Down syndrome patients. Note characteristic features consisting of upslanting palpebral fissures, epicanthal folds, a hypoplastic midface, and prominent lips. B, lateral view showing flattened anteroposterior diameter of the skull and the decreased vertical dimension of the midface. Note low nasal dorsum, decreased mentum prominence (micrognathia), and small auricle (microtia). (*Kanamori et al; 2000*)

crease is usually noted. Down syndrome may be associated with congenital heart diseases mainly septal defects. Down syndrome patient may show decreased acetabular and iliac angles, small penis and cryptorchidism. Other manifestations like short neck, small teeth, furrowed tongue, strabismus, intestinal atresia and imperforated anus are frequently seen. (*Richard and Victor, 1983*)

Recent advances in surgery for the treatment of congenital heart defects have greatly enhanced the survival of children with Down syndrome. (*Yang et al, 2002*)

However, despite this enhancement in life expectancy, the ability of children with Down syndrome to reach their full developmental potential may be diminished by disorders of the ear, nose and throat. Increasingly, caregivers seek therapy for these disorders to promote