Nigerian Journal of Paediatrics 1993; 20 (3): 60 - 3

Infantile Achalasia in Down's Syndrome

DA OLANREWAJU* AND JK RENNER+

Summary

Olanrewaju DA and Renner JK. Infantile Achalasia in Down's Syndrome. *Nigerian Journal of Paediatrics* **1993; 20:61.** A 3-year old girl with clinical features of Down's syndrome presented with persistent vomiting of undigested food and recurrent cough. Although her karyotype was not carried out, radiographs of the pelvis and hands revealed characteristic features of Down's syndrome. An investigative barium swallow to diagnose the cause of the vomiting showed achalasia which is a rare association with Down's syndrome.

Introduction

SINCE Down's syndrome was recognized as a clinical entity, congenital lesions in several organs have been associated with the condition. These phenotypic expressions are due to either a non-disjunction (trisomy - 21), or translocation on chromosome 15 or 21. All affected children look as if they belong to a single family. The radiograph of the pelvis, hip joint, skull and facial bones of patients with Down's syndrome are quite characteristic. [1,2] Intestinal congenital lesions in children with Down's syndrome have been described and these include duodenal atresia [3] and Hirschsprung's disease.[4] To the best of our knowledge, there has been no reported case of infantile achalasia in Down's syndrome. The purpose of this communication is therefore, to document this rare association.

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Case Report

N1, a three-year old female, was diagnosed clinically to have Down's syndrome shortly before the age of one year. She was delivered at term by a 34-year old mother. The birthweight was 2.65kg. She remained in hospital for two days for management of aspiration pneumonia which developed at delivery. At about the age of eight months, symptoms of persistent vomiting of both solid foods and fluids started. It was reported by the mother that undigested food taken six to eight hours previously was usually present in the vomitus. There was also coughing and these two symptoms necessitated her first admission into a hospital for 17 days. At the age of three years, she was referred to a respiratory clinic on account of persistent cough which was unresponsive to the treatment that was given. Physical examination showed a placid child with odd *facies*, weighing 11.7kg.Other characteristic features included generalized hypotonia, medial epicanthic folds, slanted eyes, protrud-

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