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Infantile Achalasia in Down's Syndrome

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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-

ing tongue and low-set ears. She had simian creases in the palms. Both motor and mental retardation were present. The cardiovascular system was normal.

Investigations carried out included barium swallow which revealed a tapering (bird's beak) of the distal oesophagus below the diaphragamtic level and dilatation of the upper oesophagus (Fig 1). Pelvic radiograph obtained in neutral position (Fig 2) showed a small acetabular angle (13.5^0), normal iliac angles (64^0) and iliac index of 77.5^0. There were coxa valga and an inwardly curving fifth finger (Fig 3). Chest radiograph showed perivascular densities in the right lung field, compatible with interstitial pneumonia. Facilities for karyotype study were not available.

Fig 1

Barium swallow showing symmetrical tapering of the oesophagus (bird's beak) at the obstruction site which is bellow the diaphragmatic level, with proximal dilatation above the obstruction site.

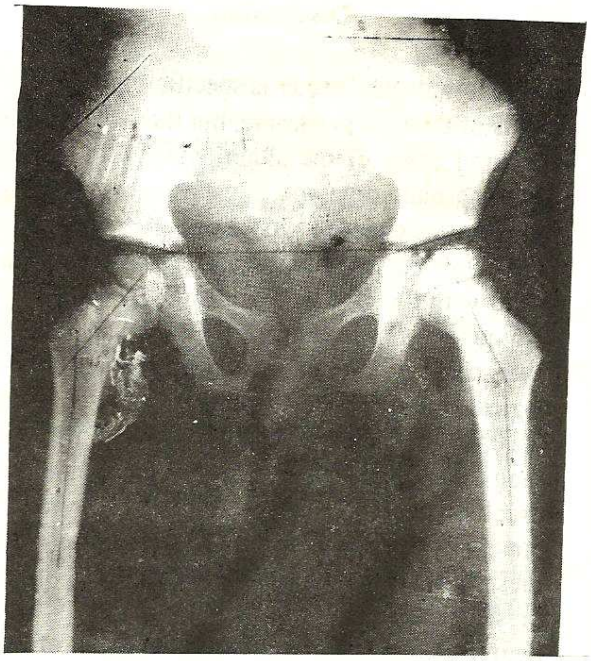

Fig 2

Frontal view of the radiograph of the pelvis showing decreased acetabular angle (R-11^0) (L-16^0) and coxa valga (R-145^0 L-155^0).

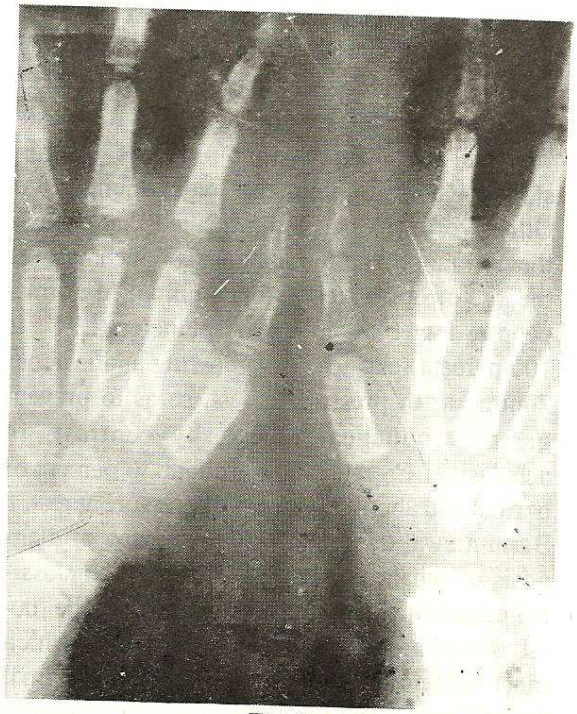

Fig 3

Radiograph of the hands showing ulnar deviation and clinodactyly, more marked on the right than on the left.

Discussion

Chromosomal study is specific for the diagnosis of Down's syndrome, but there have been reported cases of the abnormality with apparently normal karyotype, [4] just as autosomal trisomy may exist with syndromes other than classical Down's syndrome.[5] Since it is rare for a child with the clinical features of Down's syndrome to have a normal karyotype [4] which however, was not undertaken in our patient, radiological studies were performed so as to confirm the clinical diagnosis of Down's syndrome. Osseous manifestations of Down's syndrome are usually assessed before the age of one year, [12] but as Roberts *et al* [6] have reported, some of these features persist into adult life. Thus, in the present case, a small acetabular angle of 13.5 degrees, coxa valga with ulnar deviation of the hands and clinodactyly (inward curving of fifth finger) persisted at the age of three years. An iliac index of 77.5 degrees in our patient is in agreement with the finding of others.[2]

There can be no doubt that in the present case, there was considerable delay in the diagnosis of achalasia at the age of three years, whereas significant symptom of the disorder started when the child was eight months old. Other workers [7-9] have commented that even in the normal population, diagnosis of achalasia at the age of our patient is uncommon. In normal people, lower oesophageal sphincter (LES) is 100 percent relaxed on swallowing and is about 20mm Hg of pressure at rest, so as to prevent gastrooesophageal reflux; by contrast, in achalasia, LES relaxation is about 30 percent of normal and about 50mm Hg of pressure at rest.[10] Barium swallow remains the mainstay of diagnosis of achalasia of the oesophagus and classical cases show symmetrical tapering of the distal oesophagus at the obstruction site which must be below the diaphragm level,[11] proximal oesophageal dilatation above the obstruction site, and low amplitude contraction waves or aperistalsis in the body of the oesophagus. Oesophageal dilatation of a degree to produce right cardiac border image on chest films may alert the discerning radiologist to the possibility of achalasia. However, this stage of progression in our patient had not been reached. Strictures simulating achalasia, may be differentiated by valsalva manouvers [7] which will relax achalasia, but not a stricture.

Pulmonary complications of achalasia must be promptly recognised so as to prevent irreversible pulmonary damage. These complications are reportedly higher in children than in adults [8 12] However, in the series reported by Berguist *et al,* [7] 15 percent of the patients had pulmonary infection. As patients with Down's syndrome reportedly have increased frequency of respiratory infection, the interstitial pneumonia in our patient may be a summation of this factor and the achalasia.

Achalasia in childhood probably has genetic and familial factors. Allgrove *et al*[13] have described the Triple A syndrome of achalasia, alacrima and adrenocorticotrophic hormone (ACTH) insensitivity in children and they were inclined to believe in an autosomal recessive mode of inheritance. Freiling *et al* [14] have noted the rarity of familial cases and the possibility of horizontal transmission, thus, implying a similar mode of inheritance as suggested by Allgrove *et al.*[13]

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