

Down's Syndrome in Nigerian Children*

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It has been suggested in the past that Down's syndrome is rare or even non-existent among Africans. Tooth (1950) did not see any such patients during his investigation of mental illness in Ghana. Jelliffe (1954a, b) states that he did not see any children with Down's syndrome during a period of six years in the Sudan and Nigeria. He described 13 patients with the syndrome in Jamaica but thought that the defect might have been derived genetically from non-African sources.

Since then several patients and groups of patients have been described from Africa, e.g. Luder and Musoke (1955) who described 5 patients seen in Uganda; Leather and Leather (1957) with one patient; and Hassan (1962) with 30 patients seen in the Sudan. Most paediatricians working in Africa now agree that the condition is not a rare one though its actual incidence is not known.

The present paper describes only two patients, but publication is thought to be justified since the diagnosis in both was confirmed by chromosome analysis and so far as the writer is aware this is the first occasion on which this technique has been used in tropical Africa.

Case Reports

Case 1. This girl was seen for the first time on November 12, 1962 when she was 18 months old, the principal complaints being failure of growth and fretfulness. Her mother said that she was much smaller than other children of her age and quite different from all her sibs, in a way she could not understand.

She was born after a normal delivery at home, and had been fed on maize pap with some milk. She had had no significant illnesses.

The mother's age was estimated to be about 40. She had 4 other living children and had had 3 others that had died.

On examination the child weighed 9 lb. 4 oz. (4.2 kg.). Her general appearance is shown in Fig. 1 and is typical of Down's syndrome.

The *hands* had a strongly marked transverse crease pattern. Examination of the *feet* showed wide separation of the great toes from second toes. The *heart* was not enlarged. A systolic murmur was present with maximal intensity at the left sternal border, suggestive of a patent interventricular septum. There were no abnormal physical signs in the *lungs*. Examination of the *abdomen* revealed the liver edge palpable about 1 cm. below the costal margin. Radiograph of the *pelvis* showed an acetabular angle of 10 degrees.

Case 2. She was first seen on October 18, 1962, aged 8 months with the complaints that she was unable to sit up without support, had always been very snuffly, and was losing weight. Pregnancy and delivery had been normal and she had had no illnesses.

The mother's age was estimated at about 43 years. She had only one other living child, but four had died. None had resembled this one.

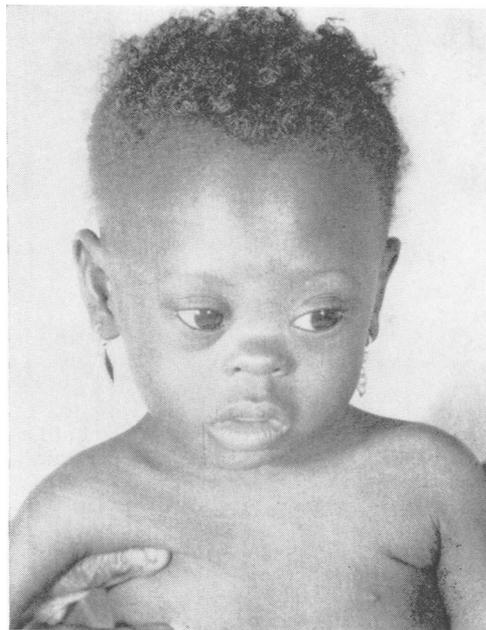


FIG. 1. Appearance of Case 1.

* Received September 18, 1963.

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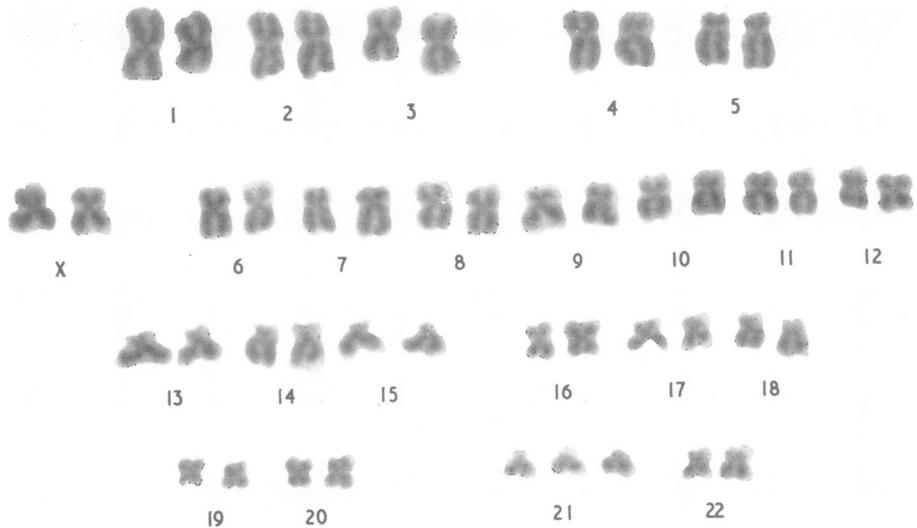


FIG. 2. Karyotypes of Case 1.

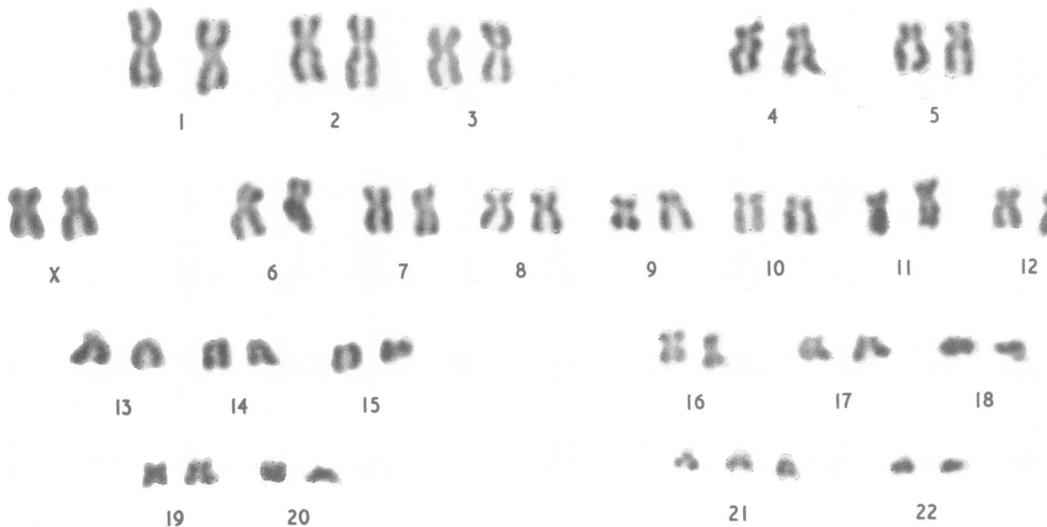


FIG. 3. Karyotypes of Case 2.

On examination the child weighed 19 lb. 10 oz. (8.9 kg.). Her facial appearance was typical of Down's syndrome. There were well-marked epicanthic folds, and a fairly well-developed occipital bulge. She had severe nasal obstruction, but no external discharge. Her hands had strongly marked transverse creases. No abnormalities of the heart were found. Examination of the lungs revealed severe inspiratory rib recession, but no other abnormal physical signs. The liver was about 1 cm. below the costal margin. A radiograph of the pelvis showed, 'acetabular angles reduced; appearances typical of mongolism.'

Chromosome Studies

Chromosome studies were carried out on white cells cultured from the peripheral blood by the Galton Laboratory modification of the method described by Moorhead, Nowell, Mellman, Battips, and Hungerford (1960). In Case 1 counts were carried out on 55 cells, and in Case 2 on 59.

The results are shown in the Table.

The karyotypes of Cases 1 and 2 are shown in Fig. 2 and 3, respectively. They both show trisomy of chromosome 21, which is characteristic of 'regular' mongols, i.e. those in whom the chromosomal abnormality is

TABLE

	No. of Cells with 45, 46, 47, 48, 49 Chromosomes				
	45	46	47	48	49
Case 1	0	3	47	3	2
Case 2	1	11	41	6	0

the presence of an extra discrete chromosome as distinct from 'translocation' mongols in whom the extra chromatin is attached to one of the other autosomes (Hamerton, 1962).

Summary

Two cases of Down's syndrome in African children are described. Chromosome analysis showed both of them to be typical 'regular' mongols with trisomy of chromosome 21.

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