TRISOMY 22: A CASE REPORT

MAJOR M. A. TETTENBORN, M.B., Ch.B., M.R.C.P., D.Obst.R.C.O.G.,
D.C.H., R.A.M.C.

British Military Hospital, Rinteln

SUMMARY: A case of Trisomy of chromosome 22 is presented. This case is of particular interest because of a partial deletion of one of the 22 chromosomes in both the patient and her mother.

The typical phenotype of the condition is discussed.

Introduction

Since the introduction of Giemsa banding allowed the adequate differentiation of Chromosomes 21 and 22 there has been a small number of reports of Trisomy 22 with a common phenotype. I wish to present one further case which is of special interest in that it is associated with a maternal chromosome abnormality.

Propositus

The propositus (Fig. 1), a female, is the first live infant born of non-consanguineous caucasian parents aged 22 and 23. Birth weight was 2840 g and the condition at birth was satisfactory. The mother had previously suffered two first trimester abortions.

Fig. 1. The propositus

At delivery the infant was noted to have a mid-line cleft palate, an accessory auricle on the left and skin tags related to both ears. There was a single palmar crease on the left hand. No other abnormality was detected at that stage, but a chromosome analysis was felt to be justified.

The baby remained well apart from a history of cyanotic spells when crying. At six weeks the infant's facies showed some mid-line prominence, the head circumference was falling below the 10th centile and there was a very minor micrognathia. The following investigations were performed:—

Chest X-ray showed apparent herniation of lung through the apex of the right thorax, E.C.G. and E.E.G. were both normal, X-ray of pelvis showed bilateral dislocation of the hips. No ocular abnormality was found by an ophthalmologist.

The hips were reduced under general anaesthetic with considerable difficulty. It was thought that they had been fixed out of the acetabulum at the time of birth.

By 12 weeks she was under the 3rd centile for both weight and head circumference. She had a development quotient of about 50 and was moderately hypotonic. The episodes of cyanosis had ceased and the chest X-ray was now normal.

By 22 weeks her weight was only 4800 g and her head circumference only 38 cms. She continued to develop slowly and her hypotonia was much less marked. Physically she remained well and her hearing and vision appeared normal.

Chromosome analysis

In the propositus 47 chromosomes were found in each of 25 cells analysed, the extra chromosome was found, with the aid of Giemsa banding, to be a 22 chromosome with deletion of part of the long arm (47XX+22q—).

In the father a normal male karyotype was found, but in the mother there was present a deletion of part of the long arm of one of the 22 chromosomes (46XX22q—). It was not possible to locate the missing fragment of the 22 chromosome thus leaving the possibilities that this represents either a balanced translocation or a simple deletion (Fig. 2).

Fig. 2. Mother's chromosome pattern

Discussion

This baby presents many of the typical features of Trisomy 22 as described in Table I. There would seem little doubt that this is a definite, if uncommon, entity.

Table I
Abnormalities found in previously reported cases of Trisomy 22

	*14 Proven cases	**11 Suspected cases
Low birth weight	6	2
Growth retardation	11	10
Mental retardation	11	10
Congenital heart defect	12	5
Elongated philtrum	5	5
Mid-line facial hypertrophy	6	4
Micrognathia	10	6
Pre-auricular appendages/sinuses	10	3
Low set ears	10	7
Anti-mongoloid slant of eyes	6	2
Mid-line cleft palate	9	4
Congenital dislocation of the hip	4	1
Finger-like thumb/hyperextensible fingers	6	3
Thoracic abnormalities	3	3
Imperforate anus	1	0
Microcephaly	11	5
Low set nipples	4	2
Hypotonia	6	7
Inguinal herniae	3	0

Notes: *Three cases died in early infancy. **One case died in early infancy.

It is of particular interest in that it is only the third case (Zackai et al 1973, Zellweger et al 1975) in which an abnormality of the extra chromosome has been found in association with a similar abnormality in the mother. This would lend support to the hypothesis that partial deletion of a chromosome will interfere with the normal processes of dysjunction.

It would now seem reasonable to offer pre-natal diagnostic procedures to parents in whom there is known to be a partial deletion of a chromosome. "Clinicians may tend to dismiss pre-auricular appendages/sinuses as common and of little significance, but this finding may be a useful reminder to search for other features of Trisomy 22, which may prove to be less rare than the small number of reported cases suggests".

It remains true that the presence of one congenital abnormality, albeit a relatively minor one such as pre-auricular appendages/sinuses, should always lead to a search for other abnormalities that may be part of a syndrome such as Trisomy 22.

It should also be borne in mind that a mid-line cleft palate carries a high association with other congenital anomalies.

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T.A.V.R. Adviser to the D.G.A.M.S.

Colonel D. C. Wilkins, M.B., B.S., D.Obst.R.C.O.G., has been selected for appointment as Territorial Army Volunteer Reserve Adviser to the Director-General of Army Medical Services, in the local rank of Brigadier, as from 1 April 1979, in succession to L/Brigadier T. W. A. Glenister, on completion of tenure.

Honorary Consultant

Dr. M. N. Maisey, M.D., F.R.C.P., has been appointed Honorary Consultant in Nuclear Medicine to the Queen Elizabeth Military Hospital, with effect from 17 April 1978. This is a new appointment.