11q Aneuploidy:
Partial Monosomy and Trisomy in the Children
of a Mother with a t(3;11)(p27;q23) Translocation

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Summary. A woman with a balanced translocation t(3;11)(p27;q23) has had
three abnormal children. The first child died in infancy, and of the two
survivors who show segregation of the derivative maternal translocated
chromosomes, one exhibits partial trisomy 11q and the other partial mono-
somy 11q. The two cases are compared with each other and with reported
examples. Moreover, 11q break points are discussed.

Introduction

Fourteen cases of partial trisomy 11q and nine of partial monosomy 11q are
reported in the literature. Of the trisomic individuals, nine were the result of
balanced maternal translocations (Francke, 1972; Jacobsen et al., 1973; Aurias et
al., 1975; Giraud et al., 1975; Laurent et al., 1975; Ayraud et al., 1976; Noel et al.,
1976), four of balanced paternal translocations (Rott et al., 1972; Tusques et al.,
1972; Wright et al., 1974), and one occurred de novo (Dinno et al., 1974). In
contrast, only one out of ten cases of partial monosomy was associated with a
paternal translocation (Jacobsen et al., 1973), the remaining nine were all
spontaneous events (Faust et al., 1974; Linarelli et al., 1975; Taillemite et al.,
1975; Turleau et al., 1975; Engel et al., 1976; Larson et al., 1976; Kaffe et al., 1977;
Frank and Riccardi, 1977; Valente et al., 1977).

This report describes both partial monosomy and partial trisomy 11q in sibs,
arising from a balanced maternal translocation t(3;11)(p27;q23).

Case Report

The mother's first baby was delivered by Caesarean section at term, after a threatened
miscarriage at 5 months of gestation. The female infant weighed 2693 g and was reported to
have been abnormal with a sacral dimple and a deformity of the foot, was a poor feeder, did not
respond to stimuli and suffered a cot death at 5 months of age.

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The second pregnancy was uneventful and proceeded to term when birth was induced (reason unknown), resulting in a female infant weighing 6902 g. Apart from excessive weight gain, which may have resulted from ACTH treatment, nothing was noted about her progress until she was 4 1/2 months old, when severe fits commenced. When seen at the age of 1 year, A.M. (see Fig. 1) was semi-conscious and showed the following features: obesity, frequent fits, hypotonia, weak cry, abnormal sucking movements, little response to stimuli, microcephaly, trigonocephaly, oval face, slightly upward slanting palpebral fissures, simple ear configuration, globular nose with upturned nares, prominent protruding philtrum, large downturned mouth with thick lips, high palate and tachycardia without obvious heart defect. Her upper limb joints were stiff and the arms showed spontaneous and evoked myoclonus (more marked on the left). The deep reflexes were very brisk and there was an intermittent spontaneous Babinski reflex. Head measurements were: length, 142 mm; breadth, 104 mm; circumference, 415 mm; cephalic index, 0.73.

A male infant (N.M.) was delivered by forceps after a third, full-term pregnancy. He weighed 2800 g and had a single umbilical vessel and a sacral dimple. After 2 days he was dusky and tachypnoeic with a ventriculo-septal defect. On examination at 2 months of age (Fig. 2) he was in a poor state of nutrition and was pale and dystrophic, with retarded mental and physical development. He had a normal skull (length, 134 mm; breadth, 103 mm; circumference, 308 mm; cephalic index, 0.77), an oval face with small eyes and palpebral fissures, epicanthic folds, long lashes, small low set ears, an upturned fleshy nose, high palate, retrognathia, thin lips and long philtrum. His upper arms were characteristically flexed in a "praying" position but without obvious hypertonia. There were brisk deep reflexes of the arms with positive neonatal reflexes. He responded poorly to stimulation.

**Chromosome Studies**

Chromosomes from lymphocyte cultures taken from the children and both parents were examined in standard and trypsin G-banded preparations. The