Incidence of Major Chromosome Aberrations in 12,319 Newborn Infants in Tokyo

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Summary. In order to ascertain the frequency of chromosome aberrations among newborn infants in Japan, a chromosome survey of a large number of newborn infants is in progress. A consecutive series of 12,319 newborn babies, 6382 male and 5937 female, have been screened for clinical manifestations of autosomal aberrations and for sex chromatin and sex chromosome aberrations. Chromosome studies were carried out on 694 infants with suspected chromosome aberrations. The clinically abnormal infants were screened by conventional staining, and banding techniques have been used in the part of the study performed since 1974. Of the clinically abnormal infants, 25 had abnormal karyotypes, including two males with a 47,XXY complement, one female with a 45,X complement, three male infants with a 47,XYY complement, two with trisomy 13 syndrome, three with trisomy 18 (including one case of mosaicism), eleven with Down's syndrome (including one case of mosaicism), one with B5p partial trisomy, one with cri-du-chat syndrome, and one with Y/D translocation. The overall results are comparable to those of previous population cytogenetic studies only in the autosomal trisomies and sex chromosome abnormalities and in that the observed frequencies were comparable to those found in studies in Caucasians.

Introduction

During the past decade several chromosome surveys on newborn infants in North America and Europe have been reported (Sergovitch et al., 1969; Lubs and Ruddle, 1970; Walzer and Gerald, 1972; Fridrich and Nielsen, 1973; Jacobs et al., 1974; Rochkov et al., 1974; Hamerton et al., 1975).

The few surveys that have been based on examination of chromosomes have been limited in size by the expense of the techniques (Court-Brown et al., 1966; Ratcliffe et al., 1970).

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No such surveys, however, have been carried out on newborn infants in Japan, even for specific abnormalities, except Down's syndrome. To ascertain the frequency of chromosome abnormalities among newborn infants in Japan, a survey of a large number of newborn infants is currently in progress. The present report describes the results obtained from the screening of 12,319 consecutive live births in one hospital. Chromosome studies were carried out on 694 infants with suspected autosomal aberrations. Sex chromatin was examined in the first 2054 female and 3311 male babies, and further chromosome analysis was carried out on the 52 of these babies who proved to have abnormal sex chromatin.

Materials and Methods

The survey included 12,319 consecutive live births at Y. Maternity Hospital in Tokyo between 1 July 1973 and 31 December 1977. This hospital is the largest maternity hospital in Hachioji, a western suburb of Tokyo, and its intake includes all socioeconomic classes.

The average birth rate was 220 babies per month during this period. The distribution of maternal ages at delivery, shown in Figure 1, was similar to that in the general population of Japan. Of the 12,319 babies examined, 6382 were male and 5937 female.

The infants underwent routine physical examinations at monthly intervals from birth to 12 months of age by pediatricians attached to the child health survey program of the hospital. Each baby was examined clinically during the first day of life by one of us, and any malformations were recorded. At the same time, a buccal smear was obtained for examination of both X (Ross, 1960) and Y chromatin (Pearson, 1970) as the primary screening method (Iijima and Higurashi, 1976). The number of X chromatin-positive cells in female infants during the neonatal period and that of Y chromatin-positive cells in male infants during the same period were markedly lower than those in young children or adults (Fig. 2). The incidence of Y chromatin-positive cells was low (36%) on the first day after birth and then increased daily to 46% on the fourth or fifth day of age. The incidence increased to 55% after 1 month, and remained at similar levels of 57% and 59% after 2 and 3 months respectively.

![Fig. 1. Distribution of maternal age at birth](image)