Trisomy 9p in a Girl whose Mother has a Translocation
t(9;20)(q12;p13)

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Summary. R banding of the fine structure of the chromatids has enabled us to study a new case of trisomy for the short arm of chromosome 9. The syndrome +9p was due to nondisjunction of a maternal translocation t(9;20)(q12;p13).

The characteristic features of the trisomy syndrome for the short arm of chromosome 9 (9p) became well known after the work of Réthoré et al. (1970, 1973). The syndrome can be pure or connected with other gene disorders. For the first time we have been able to observe a small girl whose malady originated from a maternal translocation t(9;20)(q12;p13).

Case Report

Céline was born on July 7th, 1973, after a normal, uncomplicated pregnancy, during which the mother suffered from no illness or rash of any kind, took no medicine, and was not subjected to X-ray treatment. Both father and mother were in good health. The couple had three other children, all girls, aged 11, 10, and 7 when Céline was born. These three girls and their parents, aged 36 and 31, were in good health and showed no phenotype abnormality, their intelligence being quite normal. The mother had never had a miscarriage. Delivery was normal. Céline’s weight at birth was 2930 g and the infant cried immediately.

Céline was sent to us at 10½ months because of mental retardment. Dysmorphy was immediately striking upon examination (Fig. 1). The skull was small (C.P. = 42 cm, normal

Fig. 1. Céline at the age of 10½ months
Fig. 2. Céline's karyotype