A Case of 47,XX,(21q-)+ with some Stigmata of Down's Syndrome and an IQ of 77*

RICHARD L. NEU, MARY L. VOORHESS,† and LYTT I. GARDNER

From the Genetic and Endocrine Unit, Department of Pediatrics, State University of New York, Upstate Medical Center, Syracuse, New York, USA

There are 4 cases in the literature of Down's syndrome associated with what is probably an extra 21q- chromosome (Ilbery, Lee, and Winn, 1961; Dent, Edwards, and Delhanty, 1963; Hall, 1963). This report presents another such case, with an unusual IQ of 77.

Case Report

The *proposita* was first seen at age 5 years 4 months for evaluation of possible Down's syndrome. She was the product of an uncomplicated full-term pregnancy, and the birth weight was 3920 g. The mother was 34 and the father 39 years of age at the time of the patient's birth; there are three older, healthy sibs. The *proposita* sat alone at 6 months of age, walked at 12 months, but did not begin to talk until age two and a half years. Her general health has always been good.

At examination, the patient (Fig. 1) appeared to be a pleasant, cooperative child who seemed to understand commands; her speech was quite unintelligible. Brachycephaly and hypotonia were present. The eyes had a mild mongoloid slant without Brushfield spots. The epicanthi were not prominent and the tongue did not protrude. No heart murmurs were heard. The 5th toes were short and curved in; the hands were short and broad. A dermatoglyphic study revealed ulnar loops on all 10 digits and a centrally located palmar triradius on both hands. These are both characteristic findings associated with Down's syndrome. No simian creases were present. The diagnosis of Down's syndrome was made. The patient's IQ on the Stanford-Binet scale was 77 (mental age 4 years 7 months) at 5 years 9 months.

The *proposita* has always lived at home and begun attending school at age 6 in a special, ungraded course. Menarche occurred at 12 years. The patient took the California Achievement Test at 13 years 9 months, and her grade placement was 5 (reading vocabulary 5.4, reading comprehension 5.2, arithmetic reasoning 5.4,

FIG. 1. The *proposita* at 5 years 4 months.

arithmetic fundamentals 4.9, spelling 5.4). At age 13 years 2 months her height was in the 25th centile (152 cm) and her weight was in 90th centile (64.7 kg).

Cytogenetic Studies. Leucocyte cultures were initiated for chromosome studies using a micromethod. An extra, small, abnormal chromosome (Fig. 2) was present in all the metaphases examined. The karyotype was designated as 47,XX,(21q-)+ because the clinical findings described above led to a diagnosis of

Received 15 January 1971.

* This study was aided by grant AM-02504 from the National Institute of Arthritis and Metabolic Diseases, National Institutes of Health, US Public Health Service.

† In receipt of a Research Career Program Award CA-K3-14,289, National Cancer Institute.

FIG. 2. The 21q- and other G-group chromosomes from 10 metaphases.

Down's syndrome. The abnormal chromosome was occasionally observed in satellite associations with other acrocentric chromosomes, which indicates it is derived from an acrocentric chromosome itself. The karyotypes of the parents were normal.

Discussion

One might expect that the number of clinical signs characteristic of Down's syndrome would be fewer or less apparent if much of the third chromosome 21 was deleted. The extra chromosome present in this case has most of the long arm missing, and the stigmata associated with the syndrome are diminished in degree and number. Four physicians, however, still diagnosed the *proposita* as having Down's syndrome on clinical grounds.

Minimal, but definite signs of Down's syndrome were also described for two of the other cases (Ilbery *et al*, 1961; Dent *et al*, 1963). They included Brushfield spots and a slightly protuberant tongue, neither of which was present in our case. The two cases of Down's syndrome described by Hall (1963) are somewhat confusing to interpret because the father of each patient also carried the abnormal chromosome in an otherwise normal chromosome complement. According to Hall, both patients had many signs of Down's syndrome, including heart defects; the fathers were presumably normal.

Reports concerning the intelligence of individuals with Down's syndrome vary because of bias in the selection procedures (Penrose and Smith, 1966). Wunsch (1957) measured the IQ of 77 patients with the syndrome who were being seen in a clinic for

retarded children. These children lived at home as did the *proposita* in this report; the median IQ was 38 and the highest score attained was 64. The IQ of our patient is obviously higher than would be expected, and could be attributable to the fact that she is only partially trisomic for chromosome 21.

Summary

A girl aged 13 years 2 months is described with a number of the stigmata associated with Down's syndrome, and a karyotype that has been designated as 47,XX,(21q-)+. The clinical findings are: retardation, brachycephaly, hypotonia, mild epicanthus, a slight mongoloid slant of the eyes, short broad hands, ulnar loops on all digits, and a centrally-located palmar triradius *t* on both hands. Her IQ at age 5 years 9 months was 77.

We are grateful to Dr Neil Stewart for referring the patient to our unit, to Dr Leonard Hersher who administered the Stanford-Binet test, and to Miss Saddle King for expert technical assistance.

REFERENCES

- Dent, T., Edwards, J. H., and Delhanty, J. D. A. (1963). A partial mongol. *Lancet*, **2**, 484-487.
- Hall, B. (1963). Down's syndrome (mongolism) with a morphological Philadelphia chromosome. *Lancet*, **1**, 558.
- Ilbery, P. L. T., Lee, C. W. G., and Winn, S. M. (1961). Incomplete trisomy in a mongoloid child exhibiting minimal stigmata. *Medical Journal of Australia*, **2**, 182-184.
- Penrose, L. S., and Smith, G. F. (1966). *Down's Anomaly*, p. 48. Churchill, London.
- Wunsch, W. L. (1957). Some characteristics of mongoloids evaluated in a clinic for children with retarded mental development. *American Journal of Mental Deficiency*, **62**, 122-130.