Case reports

(q11;p13). The study of polymorphisms in previous
generations of the family might have helped resolve
the origin of the abnormal chromosome but this was
not possible.

In patients found to have a twin pregnancy,
amniocentesis is likely to be attempted at a later
gestation than usual. In these circumstances it
appears advisable to carry out preliminary parental
karyotyping and thus avoid the situation described.

E. JANET WATSON
AND JOHN B. SCRMIGEOUR
Department of Pathology, Royal Hospital for
Sick Children; and the Department of
Obstetrics and Gynaecology, Western General
Hospital, Edinburgh

References
Cohen, M. M., MacGillivray, M. H., Capraro, V. J., and
Journal of Medical Genetics, 10, 74-79.
inheritance of some heterozygous Robertsonian trans-
Niebuhr, E. (1972a). Dicentric and monocentric Robertsonian trans-
Niebuhr, E. (1972b). A 45,XX,5-13-dic+ karyotype in a case of
Warburton, D., Henderson, A. S., Shapiro, L. R., and Hsu,
L. Y. F. (1973). A stable human dicentric chromosome,
t dic(12;14) (p13;p13) including an intercalary satellite
region between centromeres. American Journal of Human

Requests for reprints to E. Janet Watson, Depart-
ment of Pathology, Royal Hospital for Sick Child-
ren, Edinburgh EH9 1LF.

Leprechaunism with mosaicism
46,XX/47,XX extra ring
chromosome

SUMMARY A case of leprechaunism with a
chromosomal abnormality is reported. The
patient was a female infant, born to healthy,
consanguineous young parents. Her course was
one of extreme marasmus, with death at 3
months of age. She presented the classical features
of the syndrome and chromosome mosaicism
46,XX/47,XX,+r(?)}. It was not possible to
identify the origin of the extra ring chromosome.
It is difficult to establish the role of such a
cytogenetic finding in the aetiology of the
syndrome.

This report concerns a female infant with the clinical
features of leprechaunism, whose chromosome com-
plement was mosaic. One cell line was normal
while the other included 47 chromosomes, the
additional chromosome being a small ring. This is
the first instance in which an infant with lepre-
chaunism has been found to have such abnormal
karyotype.

Case report

The patient was born at term to healthy parents who
are second cousins. The father was 30 years old when
the infant was born, the mother 26. The mother had
had five previous pregnancies. Four had been normal
offspring while one had ended in a spontaneous
abortion. The pregnancy which produced the proband
was uncomplicated. The infant's birthweight was
3750 g and the length was 51 cm. The infant was
cyanotic. The facies was grotesque, with hyper-
telorism and oblique palpebral fissures. The nasal
bridge was broad and flat, and the nostrils large. The
lower lip was thick and everted. The palate was widely
cleft. Other abnormalities included occipital promi-
ence; low set, oblique ears with obvious malfor-
malions of the lobules (Figs. 1 and 2); and extension
of the scalp hair over the forehead. In addition, fine
lanugoty is not be present in the preauricular areas,
and on the arms and back. The nipples appeared to
be normal but were low and widely spaced. The
external genitalia were prominent. The infant was
hypertonic and maintained an opisthotonic posture
lying on her side. The hips were held in a position of
adduction. Both feet revealed an equinovarus mal-
formation with the third toes overlying the fourth.
The fists were held tightly clenched. A simian crease
was present on each palm. The skin was inelastic,
forming prominent folds, and subcutaneous tissue
was severely deficient.

Laboratory determinations, including urinary
amino acid analysis, yielded normal results. X-ray
films revealed a narrow pelvis (Fig. 3), and advanced
skeletal maturation. The bone age was 6 to 9 months
when the infant was 2 months old, on the basis of the
evaluation of carpal and tarsal ossification centres.

The infant's course was one of progressive maras-
mus and she died at the age of 3 months.
Fig. 1 Front and lateral view of the patient, showing the typical grotesque facies.

Fig. 2 General appearance, showing the opisthotonic position and the contractures.

Fig. 3 X-ray film showing narrow pelvis and advanced bone age.

CYTOGENETIC FINDINGS
Lymphocytes were cultured by standard methods. (Moorhead et al., 1960). Trypsin-Giemsa staining was employed to produce chromosome banding (Seabright, 1972). Two cell lines were observed; one normal 46,XX, the other including 47 chromosomes. The accessory chromosome (Fig. 4) appeared to be a small ring, the origin of which could not be determined. It was noted in 52 of 102 lymphocytes analysed, and its morphology was variable. A buccal smear was X-chromatin positive. Karyotypes of the parents were normal.

Discussion
Leprechaunism is a rare syndrome, characterised by unusual, grotesque facies, multiple other malformations, progressive marasmus, and early death (Donohue and Uchida, 1954). Evidence suggests that it is genetically determined, and in some families appears to conform to an autosomal recessive mode of inheritance. Functional and morphological endocrine abnormalities have been described; cystic follicles of the ovaries have been prominent
findings (Patterson and Watkins, 1962; Kallo et al., 1965; Summitt and Favara, 1969).

Table 1 summarises the principal features of leprechaunism and compares the features of the case reported herein. The phenotype of our patient is strikingly similar to those of previously reported infants with leprechaunism. An unusual feature of our case was the advanced bone age detected on x-ray examination. Such a feature may be the result of early elaboration of sex hormones, which could also explain the observed enlargement of the external genitalia.

The karyotype of the patient was most unusual. Ring chromosomes of the X and of several autosomes are known to occur. However, the ring is ordinarily one of a complement of 46 chromosomes, and not present as a 47th chromosome producing partial trisomy. The only reports, to our knowledge, in which the ring chromosome constituted an extra chromosome producing partial trisomy are those of Atkins et al. (1966), and of Varela and Sternberg (1969). In neither of the patients in those reports was the phenotype reminiscent of leprechaunism.

To date very few cases of leprechaunism have been reported: in only one was a chromosomal abnormality demonstrable (Ayraud et al., 1976). If leprechaunism is a distinct entity, inherited in an autosomal recessive manner as has been proposed (Summitt, 1974; McKusick, 1971), then it is possible that the phenotype of our patient and the chromosomal abnormality are coincidental. Our patient could be homozygous for the recessive leprechaunism gene, especially in view of the fact that her parents were consanguineous. On the other hand, it seems reasonable to attribute the patient’s phenotype to the chromosomal aberration. One previous report (Summitt and Favara, 1969) has suggested that leprechaunism is an aetiologically heterogeneous ‘physical examination syndrome’. This idea is supported by the variation in the phenotypes of patients reported with leprechaunism, and the lack of a demonstrable metabolic defect common to all patients. Moreover the only case of Ayraud et al. (1976) with a chromosomal abnormality showed a karyotype quite different from our report. We believe that further evidence is required to establish this rare syndrome as a distinct entity. Reported cases should be documented with adequate cytogenetic and metabolic investigation.

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V. VENTRUTO, L. SEBASTIO, G. SEBASTIO, R. V. DE MASI, U. VOTA, L. FARINA, AND B. FESTA
Ospedali Riuniti di Napoli. Centro Sociale Regionale per i disordini citogenetici ed ereditari del metabolismo
Divisione di Pediatría Ospedale SS. Annunziata di Napoli.
Istituto di Cibernetica del CNR Arcofelice, Napoli.
References


Requests for reprints to Professor Valerio Ventruto, Centro Sociale Regionale per i Disordini Citogenetici ed Ereditari del Metabolismo, Ospedale Cardarelli, Napoli, Italy.