THE CAT-CRY SYNDROME*

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This syndrome was first described by Lejeune *et al.*[1] in 1963 and was given the name of *Maladie du cri du chat*. Since then there have been a number of other reports of the condition, which have been well summarized in an article by Kajii *et al.*[2] in which the chief clinical features in 12 reported cases, as well as one of their own, are set out.

The characteristic features are the abnormal cry, likened to the mewing of a cat, and a persistent failure to thrive. The diagnosis is confirmed by chromosome studies which show deletion of the short arm of the fourth or fifth chromosome.

Other findings[2] may be low birthweight, an antimongoloid slant of the eyes, epicanthic folds, hypertelorism, strabismus and optic atrophy. The head may be micro- or brachycephalic with 'mooning' of the face, low-set ears, micro- or retrognathia and a short neck. Additional features described are congenital heart disease, hypotonia and abnormal dermatoglyphics.

We report a case of this syndrome—as far as we are aware for the first time in this country—partly because of its comparative rarity and also because the baby was extensively investigated for his failure to thrive. All this turned out to have been quite unnecessary, as once the diagnosis of *cri du chat* was thought of, an adequate explanation of the infant's physical abnormalities was available.

CASE REPORT

P.J., a Coloured male infant of 5 months, was admitted on 25 February 1967 after failure to respond to outpatient treatment for severe gastro-enteritis. He had been delivered by caesarean section following a severe antepartum haemorrhage. Birthweight was 5 lb. The mother claimed that despite an adequate diet he had never thrived, and at the age of 3 months he weighed only 6½ lb.

Examination showed a marasmic (6 lb.), dehydrated infant with a small head, the circumference measuring 13⅝ inches. He had bilateral inguinal hernias, a 2-finger enlargement of the liver and a systolic bruit at the left sternal border. He was also noticed to have epicanthic folds, a receding jaw and a 'mewing' cry. The nursing staff called him 'the cat'.

There was consolidation of the right upper lobe on the chest X-ray film, and an intravenous pyelogram indicated incomplete rotation of the left kidney. An air encephalogram suggested bilateral cortical atrophy, especially on the left (Fig. 1).

Chromosomal Analysis

This was performed on peripheral blood lymphocytes using a modification of the technique of Moorhead *et al.*[3] The nodal number was 46 and the sex chromosomal complement XY. Eleven karyotypes were studied and in all of them the short arm of the B group showed partial deletion. The quality of the preparation, however, made it difficult to decide whether the deleted chromosome was number 4 or 5.

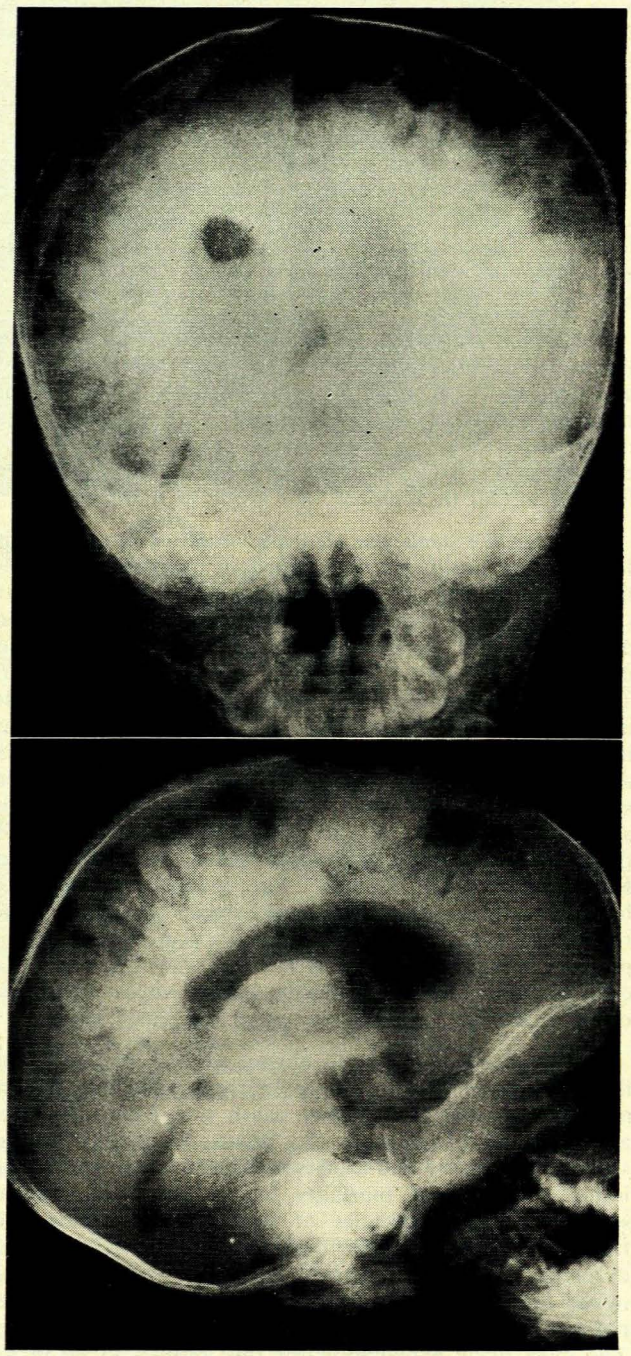

Fig. 1. Air encephalogram showing cortical atrophy.

*Date received: 11 April 1968.

The baby remained for a long time in hospital, requiring frequent intravenous infusions of electrolytes as well as plasma and blood. Correction of metabolic acidosis with sodium bicarbonate was required on several occasions. Despite penicillin and sulphadiazine the radiological signs of consolidation persisted, while the diarrhoea remained troublesome, resisting alterations in the diet, such as the removal of lactose. Ultimately the loose stools came under control, there was some gain in weight and the child was discharged from hospital nearly 4 months after his admission.

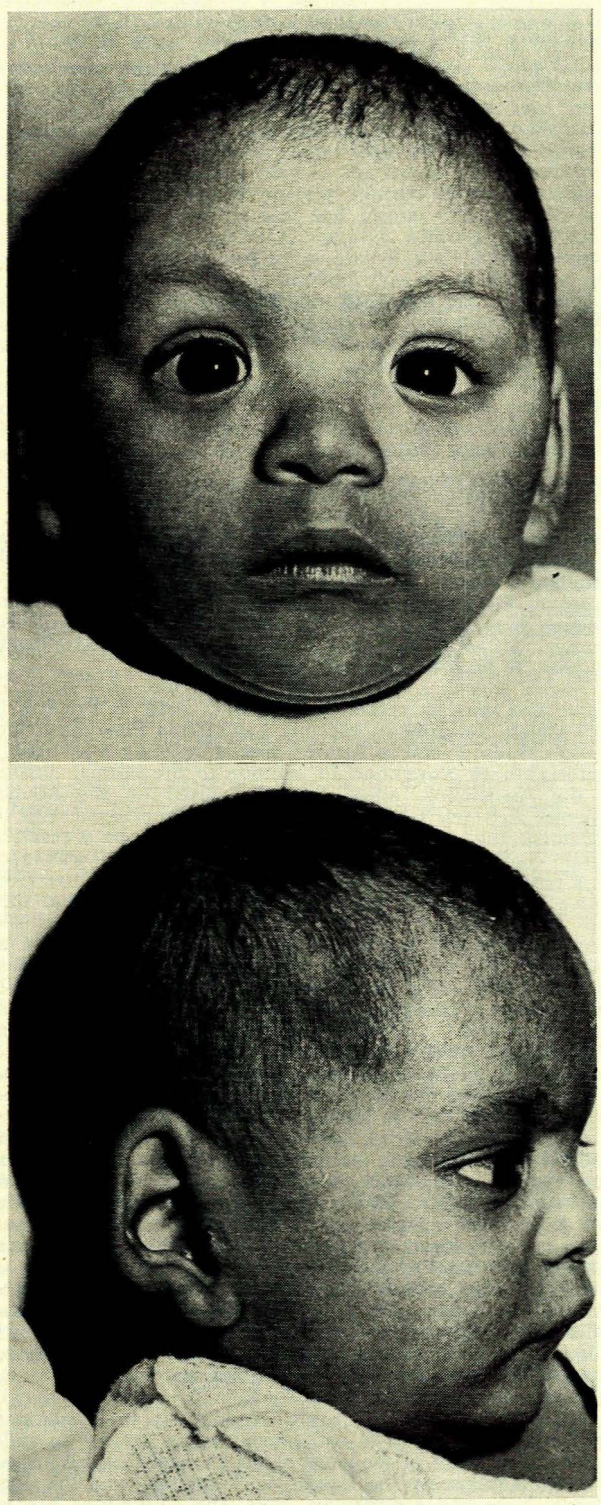

Fig. 2. P.J., aged 18 months. Note 'mooning' of face, epicanthic folds, hypertelorism and underdeveloped mandible.

He was seen again at the age of 18 months. His face had become rounded but he could not sit, or even hold up his head. He was obviously severely retarded mentally. The cat-like cry was still present. Photographs taken at this time show the typical facies of the syndrome (Fig. 2). The opportunity was taken to do a laryngoscopy, and a small larynx, with some sucking in of the epiglottic structures during forced inspiration, was reported. These findings were not regarded as significant.

DISCUSSION

Our patient appears to be a typical example of the 'cat-cry' syndrome. He has the characteristic cry, with failure to thrive and abnormality of chromosomes as well as some of the clinical features described by others, such as microcephaly, epicanthic folds, moon facies and receding jaw. The importance of the condition, although rare, is that it can be diagnosed soon after birth. This will save many unnecessary investigations such as were carried out on this patient before the correct diagnosis was suggested.

The abnormal cry of these infants has been studied by Vuorenkoski *et al.*[4] in 8 affected children. They showed that the cry, as analysed by the sound spectograph, is typical and specific for the syndrome, and quite different from the cry of normal children. Interestingly enough, there were close similarities between the *cri du chat* signals and the cries of young cats. Thus the title given to this curious syndrome is scientifically justifiable.

The anatomical basis for the abnormal cry has not been established. MacIntyre *et al.*[5] recorded the following abnormalities in some of the reported cases: laryngomalacia, an abnormally small larynx and a small epiglottis. Not all of these were present in the same child, and in some no abnormality was detected. In our patient there was no gross laryngeal disorder present.

Hobolth and Mikkelsen[6] quoted the oldest living child with the condition as being 13 years. Prognosis, not only for mental development, but even for survival, is poor. Our patient exhibits considerable mental and physical retardation. The additional findings of cortical atrophy, inguinal hernia and an abnormal pyelogram may or may not be related to the syndrome. They are not mentioned in the literature reviewed, but as more cases are published more abnormalities may well come to light. The chromosomal aberration may be due to translocation in one of the parents.[6] This aspect could not be pursued in our case as the father of the child is not living.

SUMMARY

An infant with the cat-cry syndrome is reported, showing the classical features of a cat-like cry, failure to thrive and deletion of the short arm of the B group of chromosomes. Some of the other described abnormalities were also present as well as others not previously mentioned; it is not certain whether these are of significance or not.

Awareness of the condition should prevent unnecessary and extensive investigations, as the abnormality is readily detectable and the diagnosis can be confirmed by chromosomal studies.

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