GAUCHER'S DISEASE

Report of Two Cases*

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Gaucher's disease is a rare heredofamilial disorder of cerebroside metabolism and is characterised by splenomegaly, pigmentation, pingueculae and an insidious course in adults. The course in infants, unlike that in the adult form, is very acute and often rapidly fatal.

This disease has been reported only twice[1,2] in Turkish medical literature, and for this reason it is thought to be rare in this country. We felt therefore that the appearance of two cases in a year at the Hacettepe Children's Hospital was of interest. The first case is of further interest in that it is the only infantile case reported in Turkey. The absence of neurologic findings in this case also seems to be worth recording.

Case Reports

Case 1: This was an 11 month old white boy who was admitted to the hospital with complaints of fever, cough, pallor, irritability and weakness. His mother reported that the baby was in good health until one month prior to admission, when he began to cough and developed anorexia. He was given penicillin injections by his family doctor, which relieved the symptoms of cough and brought the temperature down to normal. However, the baby continued to have anorexia and lost weight.

Past History: This was the fifth child of this family who was born after an uncomplicated pregnancy and delivery. Birth weight was unknown.

Development: The baby had head control when he was two months of age and had just begun to sit up at the time of admission. He had no teeth on admission.

Family History: His father, mother and four siblings were in good health. Bone marrow aspiration of the father and two siblings showed no abnormality.

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Physical examination: Generally this was a poorly developed, poorly nourished 11 month old white male who was pale and irritable and seemed to be chronically ill. His weight on admission was 6000 grams, length 65 cm., head circumference 42.5 cm. Pertinent physical findings included hepatosplenomegaly with the liver 5 cm. below the right costal margin and spleen down to the iliac fossa. There were enlarged lymph nodes, up to 2 cm. in diameter, in the occipital anterior cervical and axillary regions. A few rhonchi were heard in both lung fields and a grade I systolic murmur was heard over the precordium.

Laboratory Findings: X-ray examination of the lungs revealed peribronchial infiltration over the right apex. A long bone survey and intravenous pyelogram were not contributory.

Blood: The hemoglobin content was 9.1 grams per cent and the white blood cells numbered 8200 per cubic millimeter. The patient was given a whole blood transfusion after the hemoglobin had dropped down to 5.1 grams per cent.

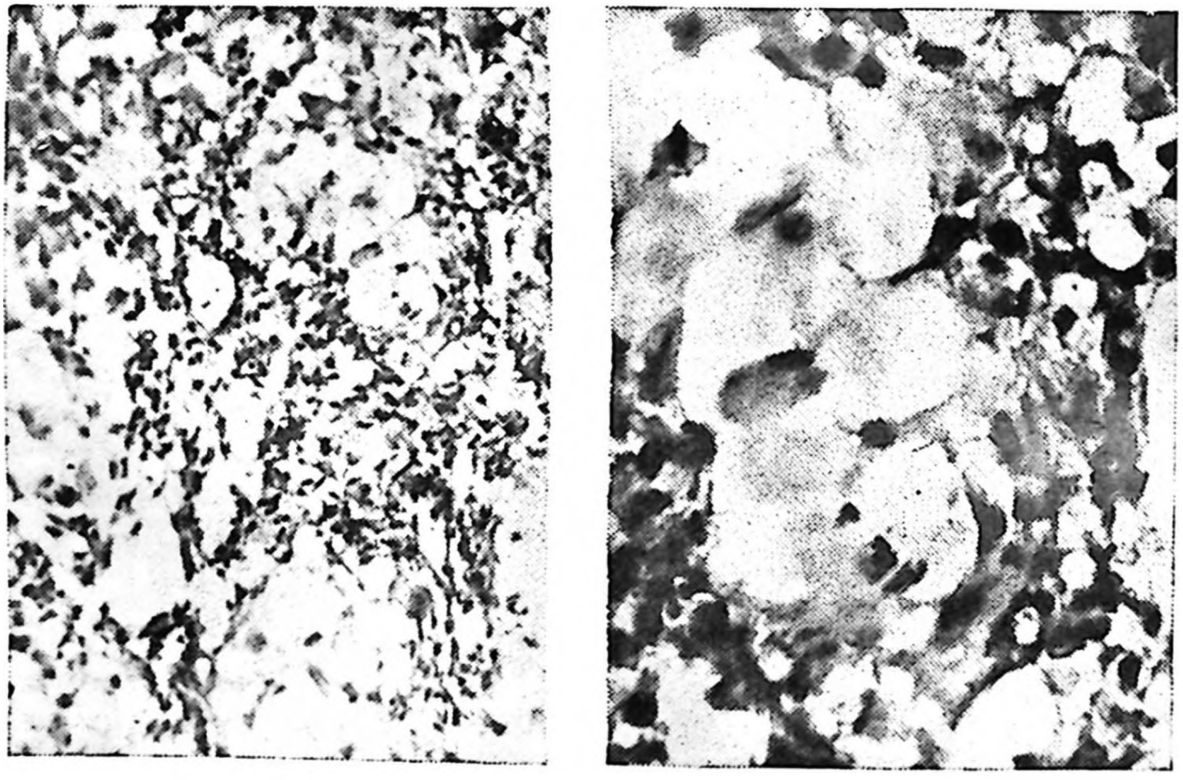

Fig. 1. — Gaucher's cells in spleen biopsy of Case 1.

No Gaucher's cells were demonstrated by bone marrow aspiration biopsy; however, biopsy of the spleen revealed abundant Gaucher's cells.

The patient showed little improvement following blood transfusions and symptomatic treatment, and was discharged after 49 days of hospitalization. He died nine months later at home.

Case 2: This was a 12 year old girl who was brought to our hospital with a history of abdominal distention and pallor, which were first not d when she was six years of age. She also complained of abdominal pain over the left upper quadrant. The patient had had epistaxis several times during the last three or four months prior to admission. She was splenectomized in another hospital, with a diagnosis of splenomegaly secondary to malaria, about ten days before her admission to the Hacettepe Childrens Hospital.

The routine laboratory findings here were not contributory except for the presence of Gaucher's cells in the bone marrow. Many such cells were reported to have been seen in the removed spleen.

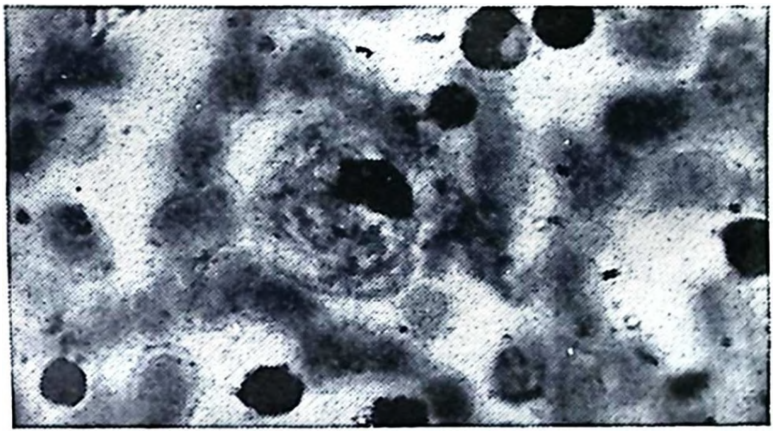

Fig. 2. — Gaucher's cells in bone marrow of Case 2.

There was no anemia on admission since she had been given whole blood during splenectomy. Her physical examination revealed that the liver was 5 cm. below the costal margin. There was pigmentation over the anterior surfaces of both forearms. No other significant findings were noted. The patient was discharged after 7 days of hospitalization. She was given no treatment during this period.

Discussion

Gaucher's disease is a rare hereditary condition which is transmitted by an autosomal dominant or recessive gene.[3] The incidence of this disease in males and females is reported to be equal.[4] It presents in two clinical forms, the adult type and the acute type. The adult form is characterised by splenomegaly, pigmentation, pingueculae and an insidious course. The symptoms of this form start insidiously before the

tenth year of age in about half the cases and before the thirtieth in the remainder. The acute form usually starts in the first six months of life and its course is progressive, resulting in death usually before the end of the second year of life. The etiology and pathog n sis of this condition are little understood. It seems that this is the result of a metabolic disturbance of the reticulum cells and is most likely due to intracellular enzymatic imbalance.[5]

Treatment is symptomatic and includes blood transfusions for anemia.

In 15 of the 29 cases studied by Medoff and Bayrd,[6] pancytop - nia was present and splenectomy was done. They report that the average life span after diagnosis was longer in the splenectomized group and suggest that splenectomy is of therapeutic value. There is no effective dietary treatment of this disease.[7]

Summary

Two cases of Gaucher's disease are reported, one of them being the acute infantile form and the other apparently the adult type. The first case had no evidence of neurologic symptoms and died under two years of age at home.

Aknowledgement

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REFERENCES

1. Egemen, M., and Yücel, F. A. : Bir Gaucher hastalığı vak'ası. Hastahane, **11** : 320, 1957.
2. Tayanç, M. M. : Bir Gaucher hastalığı vak'ası. Hastahane, **12**:66, 1958.
3. Hsia, D.Y., Naylor, J., and Bigler, J. A. : Gaucher's disease - Report of two cases in father and son. Review of the literature. New England J. Med., **261** : 164, 1959.
4. Nelson, W. E. : Textbook of pediatrics, sixth edition, Philadelphia, W. B. Saunders Co., 999, 1959.
5. Thannhauser, S. J. : Lipidoses, disease of cellular lipid metabolism, second edition, New York, Oxford University Press, 1950.
6. Medoff, A. S., and Bayrd, E. D. : Gaucher's disease in 29 cases: Hematologic complications and effect of splenectomy. Ann. Int. Med., **40**: 581, 1954.
7. Van Creveld, S. : The lipidoses, Advances in Pediatrics, IV, Chicago, Year Book Publishers Inc., 1953.