A- β -LIPOPROTEINAEMIA (BASSEN-KORNZWEIG SYNDROME)

REPORT OF A CASE

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In 1950 Bassen and Kornzweig described a new syndrome when they reported the presence of irregular erythrocytes in an 18-year-old Jewish girl who had retinitis pigmentosa and a neurological disorder similar to Friedreich's ataxia. Several years later they described the same triad (abnormal erythrocytes, retinitis pigmentosa, and neuropathy) in the patient's brother (Kornzweig and Bassen, 1957). The parents of these sibs were first cousins.

In 1952 Singer, Fisher, and Perlstein described a similar case and suggested the term acanthocytosis for the irregular red cells (Akantha = thorn). This patient had irregular red cells and neuropathy. Retinopathy, though not present at 13½ years, had developed by 19 years (Jampel and Falls, 1958). Druetz (1959) later reported a case from Belgium and suggested that the correct term was acanthocytosis. Druetz's patient had hypocholesterolaemia, a feature that had been noted in one of the previously reported cases (Jampel and Falls, 1958).

In 1960, Salt, Wolff, Lloyd, Fosbrooke, Cameron, and Hubble described a child suffering from acanthocytosis and demonstrated virtual absence of serum low-density lipoproteins (i.e. β -lipoproteins). This finding has been confirmed in other patients with this syndrome.

Twelve cases with this condition have now been reported. Steatorrhoea, hypocholesterolaemia, reduction of total serum lipids and serum phospholipids are now known to be features of the syndrome. Acanthocytosis and A- β -lipoproteinaemia occur in all cases, but neuropathy and retinopathy have not been present in all. Where biopsy of the small intestinal mucosa has been carried out the histological appearance has always been the same. The columnar cells covering the villi have unusually clear cytoplasm (Salt *et al.*, 1960; Mabry, Di George, and Auerbach, 1960; Lamy, Frézal, Polonovski, and Rey, 1961;

Schwartz, Rowland, Eder, Marks, Osserman, Hirschberg, and Anderson, 1963; Ways, Reed, and Hanahan, 1963).

The patient to be described was diagnosed at the age of 10 months and showed some unusual features that led to his early death.

Case Report

The patient, J.M., was born on April 19, 1962 following a 39-week pregnancy, his birth weight being 5 lb. 14 oz. (2,663 g.). At the age of 3 months he developed lobar pneumonia and was admitted to the Waikato Hospital under the care of Dr. D. H. H. Pullon. At the time of admission he was malnourished, his weight being 7 lb. 12 oz. (3,515 g.). Investigations at this time revealed Hb 9.2 g./100 ml. Microspherocytes, burr cells, and hypochromic macrocytes were described in the blood film. The erythrocytes showed decreased osmotic fragility. The serum non-protein nitrogen which was 31 mg./100 ml. on the first examination later rose to 64.74 mg./100 ml. The bowel motions were bulky and 53% of the dried weight was fat. A non-specific aminoaciduria was present. An intravenous pyelogram was normal. During his stay in hospital his motions were loose. No pathogens were isolated from the stools at first but later *Staphylococcus aureus* was isolated. A recurrence of pneumonia at the age of 8 months responded satisfactorily to antibiotics.

He was given a course of iron dextran intramuscularly, and folic acid, and his haemoglobin rose to 15.2 g./100 ml.

At the age of 9 months he weighed 10 lb. 12 oz. (4,875 g.) and was transferred to the Karitane Hospital, Auckland, for feeding management. At the age of 9½ months he developed basal bronchopneumonia and was transferred to the Princess Mary Hospital for Children under the care of Dr. R. H. Caughey.

Family History. The child's parents are Maoris who are second cousins. The father suffers from spinal tuberculosis. There are 8 living sibs, 3 of whom are adopted out. A ninth sib died at the age of 1 year from gastro-enteritis.

Examination. At the time of admission examination revealed a mentally retarded Maori child (mental age 4 months, chronological age 10 months). He weighed 11 lb. 5 oz. (5.2 kg.). His head appeared small and the occiput was flattened. Slight flexion contractures of the knees were present. The deep reflexes were diminished in the legs. Signs of bronchopneumonia with bronchospasm were present. There was no evidence of retinopathy.

TABLE 1
HAEMATOLOGICAL DATA

Hb (g./100 ml.)	11.6
Haematocrit (%)	36
MCHC (%)	32
Acanthocytes	35-40% of erythrocytes
Reticulocytes	2/100 RBCs
WBC	7,600/c.mm.
ESR (Westergren)	3 mm. in 1 hour
Red cell osmotic fragility	Decreased
Direct Coombs test	Negative
Plasma bilirubin (mg./100 ml.)	0.2 mg./100 ml.

Investigations. Preliminary examination of the peripheral blood revealed a haemoglobin of 11.6 g./100 ml. Approximately 35% of red cells were conspicuously small and darkly staining. Irregularity of shape of the majority of these cells was at first dismissed as artefact and the cells identified as microspherocytes. Demonstration of decreased red cell fragility led to examination of further blood films and recognition of the consistency of the red cell irregularity (Fig. 1). This irregularity was present in wet preparations, on suspension of the cells in normal saline, and was particularly well seen by phase contrast microscopy (Fig. 2). Under phase contrast the abnormal cells showed intense golden yellow coloration, contrasting sharply both with normal red cells and with cells showing normal crenation, the latter being numerous in all preparations examined. Each abnormal cell had one or more spiculate projections which frequently branched or curved and often had a length greater than the diameter

TABLE 2
LIPID AND RELATED STUDIES

Serum β -lipoprotein (mg./100 ml.)	10	3 separate estimations (normal 130-500)
Turbidometric method*	20	
	15	
Immunological assay†	None detected	
Ultracentrifugal analysis‡	Low density (< 1.063)	
	lipoproteins absent; chylomicra absent	
Serum cholesterol (mg./100 ml.)§	23 (normal 120-220)	
Serum fatty acid esters (mEq/l.)	2.5 (normal 9-14)	
Serum carotenoids (μ g./100 ml.)	12.5 (normal 60-260)	
Faecal fat excretion (g./day)	11.6	
D-xylose absorption	24% excretion in urine	
Sweat sodium (mEq/l.)	33	
Sweat chl.-ride (mEq/l.)	38	
Total serum protein (g./100 ml.)	6.3	
Electrophoretic analysis serum proteins	Slight increase γ -globulin	
Barium meal and follow through	Moderate dilatation of small bowel; thickened jejunal mucosal folds; some clumping of barium	
Radiological bone-age	Slightly retarded at 10 mth.; normal at 17 mth.	

* Method of Walton and Scott (1964).

† Method of Soothill (1962).

‡ Modification of method of Havel, Eder, and Bragdon (1955).

§ Method of Sackett (1925).

of the body of the cell, resulting in many bizarre shapes. There was no rouleaux formation.

In view of the peculiar shape of the red cells, and the clinical picture, the possibility of the child having the Bassen-Kornzweig syndrome was entertained.

Further investigations (Table 2) revealed virtual absence of serum β -lipoproteins, profound hypocholesterolaemia, considerable reduction of serum fatty acid esters and serum carotenoids. Steatorrhoea was present.

These investigations confirmed that the child was suffering from the Bassen-Kornzweig syndrome.

Further investigation (Table 3) revealed a persistently raised blood urea. A generalized aminoaciduria was present but the serum amino acids were normal, indicating that the aminoaciduria was of a low renal threshold type.

FIG. 1.—Peripheral blood film showing darkly staining acanthocytes. (Leishman. $\times 2000$.)

FIG. 2.—Red cells suspended in normal saline. (Phase contrast. $\times 1400$.) The six acanthocytes in the field show up brightly, contrasting with crenated cells.

TABLE 3
ASSESSMENT OF RENAL FUNCTION

Blood urea (mg./100 ml.)	40-67
Serum creatinine (mg./100 ml.)	1.1
Creatinine clearance (uncorrected) (ml./min.)	15
Creatinine clearance (corrected for surface area) (ml./min.)	66
Plasma CO ₂ -content (mEq/l.)	21.9
Serum phosphorus (mg./100 ml.)	5.6
Urine pH	5.4
Urine bacterial count/ml.	10,000
Urinary amino acids	Non-specific aminoaciduria
Serum amino acids	Normal

No glycosuria was present and the urine was free from protein and infection.

Investigation of Family. The blood of both parents and five of the patient's sibs was examined for acanthocytes, serum β -lipoprotein, serum cholesterol, and serum carotenoids. No abnormality was detected (Table 4). The other three sibs have been adopted out of the family and were not available for testing.

Progress. During his stay in hospital the patient had several episodes of asthma which responded satisfactorily to treatment. No dietary treatment such as fat restriction was attempted (cf. Lamy, Frézal, Polonovski, Druetz, and Rey, 1963), because it was considered that dietary management would not be continued following discharge from hospital. Vitamin supplements were given.

He was discharged from hospital at the age of 14½ months weighing 14 lb. 3 oz. (6.4 kg.). He was re-admitted for reassessment of renal function at the age of 17 months. At the time of admission he was suffering from otorrhoea and basal bronchitis for which he received antibiotics. There was still no evidence of retinopathy. Further investigations revealed blood urea 40 mg./100 ml.,

creatinine clearance 15 ml./min.; serum phosphorus 5.6 mg./100 ml., Na 144 mEq/l.; K 5 mEq/l.; plasma CO₂-content 21.9 mEq/l.; pH of urine 5.4, urine bacterial count 10,000/ml.

Three weeks after readmission to hospital he developed an exacerbation of bronchitis and asthma which was associated with cardiac enlargement and left ventricular failure. He responded initially to digoxin but deteriorated again and died on the seventh day of the illness. Unfortunately consent for necropsy was not obtained.

Discussion

The patient described was undoubtedly a case of the Bassen-Kornzweig syndrome.

Retinopathy was not present in our patient. Retinal changes have been observed in 6 of the 12 previously reported cases (Bassen and Kornzweig, 1950; Jampel and Falls, 1958; Kornzweig and Bassen, 1957; Druetz, 1959; Friedman, Cohn, Zymaris, and Goldner, 1960; Mier, Schwartz, and Boshes, 1960) and electroretinography has been abnormal in one case in which the retinoscopy was normal (Lamy *et al.*, 1961). The 5 patients without retinopathy were all under the age of 14 years at the time of reporting (Salt *et al.*, 1960; Mabry *et al.*, 1960; Wolff and Bauman, 1961; Schwartz *et al.*, 1963; Ways *et al.*, 1963). The patient reported on by Singer *et al.* had no retinopathy at the age of 13 years, but at the age of 19 marked atypical retinitis pigmentosa was present (Jampel and Falls, 1958). It is probable therefore that all patients with this disorder will develop retinopathy as they become older.

Neurological abnormalities, particularly areflexia,

TABLE 4
SERUM CHOLESTEROL, CAROTENOIDS AND β -LIPOPROTEIN, AND ACANTHOCYTOSIS IN PATIENTS AND RELATIVES

Subject	Age-group or Age	Serum Cholesterol	Serum Carotenoids	Serum β -Lipoprotein Determined as			Acanthocytes
				Protein by Gel-diffusion Immuno-assay	Lipoprotein by Dextran Sulphate Precipitation	Low Density (< 1.063) Lipoprotein by Salt Density Method of Fractionation	
Approximate normal range	0 to 10 yr.	120 to 220 mg./100 ml.	60 to 260 μ g./100 ml.	50 to 150% of normal control	120 to 570 mg./100 ml.	—	None
	30 to 40 yr.	—	—	—	250 to 780 mg./100 ml.	300 to 750 mg./100 ml.	None
Patient J.M.	10 mth.	23	12.5	0	15	0	Present
Mother	37 yr.	164	112	75	410	—	Absent
Father	38 yr.	236	122	150	800	—	Absent
Brother	17 yr.	164	132	75	375	—	Absent
Brother	15 yr.	136	99	75	360	—	Absent
Sister	11 yr.	141	167	75	360	—	Absent
Sister	8 yr.	177	177	75	440	—	Absent
Sister	5 yr.	180	—	50	240	—	Absent
Patient of Salt <i>et al.</i>	17 mth.	22	0	0	20	0	Present
Mother of Salt's patient	30 yr.	127	84	50	—	—	Absent
Father of Salt's patient	32 yr.	107	68	50	—	—	Absent

have been present in most of the 12 reported cases (Schwartz *et al.*, 1963). Our patient showed diminished tendon reflexes, and this almost certainly represented the early stages of the neuropathy of this disease.

Steatorrhoea, which was present in our patient, appears to be a constant feature in this disease. Biopsy of the small intestinal mucosa was not carried out in our patient. In contrast to coeliac disease the glucose tolerance test in patients with the Bassen-Kornzweig syndrome is usually normal (Lamy *et al.*, 1963). In several cases with this disorder the D-xylose absorption test has been normal (Mier *et al.*, 1960; Mabry *et al.*, 1960; Lamy *et al.*, 1963), but in others it has been abnormal (Schwartz *et al.*, 1963). Two oral glucose tolerance tests were carried out on our patient. The first one at the age of 11 months showed apparent decreased tolerance with the blood sugar still above fasting levels at 2 hours. The second tolerance test at 17 months was normal. The D-xylose absorption was normal.

Lipid Studies. The low serum cholesterol level fell within the range of 20-50 mg./100 ml. previously reported for this syndrome. Ultracentrifugal analysis of our patient's serum using a density gradient method showed that the small amount of cholesterol present was associated with the high density lipoprotein (α -lipoprotein). This analysis showed no detectable low density lipoprotein (i.e. β -lipoprotein), and no chylomicra were present. The lipoprotein pattern was similar to that reported by Schwartz *et al.* (1963) and Salt *et al.* (1960). The density gradient used was identical to that employed by Dr. K. W. Walton in investigations on the case reported by Salt and his colleagues.

The dextran sulphate turbidometric method for assay of β -lipoprotein was developed after Salt *et al.* had published their paper. However, the turbidometric assay was later carried out by one of us (P.J.S.) on the serum from their patient and gave a result of 20 mg./100 ml. This result, and that of 15 mg./100 ml. in our own patient, indicate virtual absence of β -lipoprotein. The small amount of turbidity recorded represents a fixed error due to precipitation of some γ -globulin components other than low density lipoproteins.

Gel diffusion precipitin analysis (Soothill, 1962) showed no detectable protein component of β -lipoprotein. The antibody used was prepared by Dr. Soothill and reactions of identity had been undertaken previously to confirm that the antiserum was specifically directed against human β -lipoprotein. Employing this particular antiserum, the method was capable of detecting β -lipoprotein concentration of

the order of 2 mg./100 ml. (whole lipoprotein molecule) or 0.4 mg./100 ml. of β -lipoprotein protein component. The results obtained in our patient were identical to those found by Dr. Soothill in Salt and Wolff's patient. Immuno-electrophoresis also failed to reveal any detectable β -lipoprotein in our patient's serum.

Conversely, we failed to produce protein precipitation lines on gel diffusion plates when we used our patient's serum as a potential source of precipitin against varying dilutions of purified β -lipoprotein in saline and against normal serum. Allotypes of human β -lipoprotein have been described (Blumberg, Bernanke, and Allison, 1962). Blumberg (1963) has observed, in the serum of two patients with the Bassen-Kornzweig syndrome, precipitins capable of reacting with β -lipoprotein of normal serum. It is not yet clear whether such precipitins are naturally occurring antibodies or the result of an immune phenomenon.

We did not undertake fatty acid or phospholipid analysis of serum or red cells (cf. Phillips, 1962). In normal people 70% of plasma cholesterol, 50% of phospholipid, and virtually all of the plasma carotenoids are carried by the β -lipoproteins. The immunological studies showing virtual absence of the protein components of β -lipoprotein in our patient are compatible with the hypothesis that one of the major defects was complete absence of this normal transport system. Schwartz *et al.* (1963), Mabry *et al.* (1960), and Ways *et al.* (1963) suggest that abnormalities in phosphatide distribution may be of more fundamental importance in this syndrome than defects in absorption or metabolism of particular lipid components such as linoleic acid.

Acanthocytosis. In young children without the ocular and neurological manifestations of the syndrome, recognition of acanthocytes in blood films is the most likely means by which the diagnosis will be reached. In our patient the red cells were recognized as abnormal in infancy, but at various times the use of the terms 'burr cell', 'pyknocyte', and 'microspherocyte' directed attention to other clinical syndromes and delayed the eventual diagnosis. For this reason some consideration of the definition of acanthocytes and their differentiation from other abnormal red cells is indicated, particularly their separation from crenated cells, 'burr cells', and 'pyknocytes', terms that, in the purely descriptive sense, might equally well be applied to the acanthocyte.

DEFINITION AND DIFFERENTIATION FROM CRENATION. The original description of the red cells by

Bassen and Kornzweig remains the most detailed and the most vivid.

'No two cells look exactly alike. In general they present a crenated appearance but of such degree that they took on bizarre shapes simulating small beetles, crabs and turtles. Others were star shaped. The variations depended on the number and length of what appeared to be appendages growing out of the cells. Some of the cells appeared small and deeply stained. They resembled spherocytes from which buds or pseudopods were protruding and these cells in particular varied from ordinary crenation.'

Singer *et al.* (1952) likewise noted that among the abnormally shaped erythrocytes many were 'relatively small and deeply stained, thus resembling crenated spherocytes'.

It is considered that the name acanthocyte is best restricted to these small deeply staining cells in which the thorny appearance is most marked. They can then be differentiated from cells identical in appearance to normal red cells crenated by hypertonic solutions. Such crenated cells, having normal size and staining intensity, appeared in considerable numbers in dry films and wet preparations of blood from our patient. The difference between acanthocytes and crenated cells was less obvious in the thinner parts of the film but was readily seen in saline suspension and was found to be accentuated using phase contrast microscopy. With phase contrast, 35 to 40% of red cells had the appearance of acanthocytes, and crenated cells were present in approximately equal numbers, proportions that are similar to those shown by illustrations accompanying other case reports. The description by Salt *et al.* of the acanthocyte as having a serrated margin with a cog-wheel appearance is more appropriate to crenation, and does not do justice to the striking variation in outline shown by the typical cells. The presence of acanthocytosis has been claimed in two cases of hereditary vitreo-retinal degeneration and one of Eales's disease (Kahán, Kahán, and Benkő, 1963a, b). The cells illustrated by these authors do not conform with the definition of the acanthocyte given above and appear to be showing crenation. Furthermore, in these cases the sedimentation rates were not depressed and serum lipid levels were not comparable with those found in the Bassen-Kornzweig syndrome.

DIFFERENTIATION FROM MICROSPHEROCYTOSIS. Confusion with microspherocytosis results from an artefact occurring in thinner parts of dry films in which the projections on the surface of the acanthocyte become less obvious and many cells appear small, darkly staining, and spherical. Examination of the thicker parts of the film is therefore essential.

DIFFERENTIATION FROM 'BURR' CELLS. Burr cell was a term introduced in 1949 by Schwartz and Motto to describe a cell '7.5 microns or less in diameter, having one to several spiny projections along its periphery'. These cells were associated with uraemia, carcinoma of the stomach, and bleeding peptic ulcers. Subsequently other authors (Dacie, Mollison, Richardson, Selwyn, and Shapiro, 1953; Allison, 1957; Aherne, 1957; Shumway and Miller, 1957; Lock and Dormandy, 1961) described similar cells and other bizarre types of poikilocyte in cases with renal failure of varied cause, but often in association with haemolytic anaemia and thrombocytopenia occurring in childhood.

Brain, Dacie, and Hourihane (1962) have suggested that the common pathogenesis is not uraemia *per se*, but disease in small blood vessels, and have suggested the term 'micro-angiopathic haemolytic anaemia' for the condition. The abnormal red cells described by Brain *et al.* included red cell fragments and various contracted and distorted microcytes of which triangular and helmet-shaped forms were common, plus crenated cells. Crenation occurring in cells already contracted and distorted was considered to give rise to cells corresponding exactly with the burr cells described by Schwartz and Motto (1949). Schwartz and Motto in fact made no mention of contraction of cells in their description; nevertheless it is quite clear that occasional cells indistinguishable from the acanthocyte will be seen in blood films in association with the 'burr cell phenomenon', 'microangiopathic haemolytic anaemia', or 'red cell fragmentation syndrome', whichever name is considered the most appropriate. No diagnostic difficulty should arise, because in the latter condition the characteristic cells will be only one of many abnormal cell types which together comprise only a minority of red cells. In contrast, in acanthocytosis, at least one-third of all red cells will have the typical appearance, and crenation will be the only other red cell abnormality.

DIFFERENTIATION FROM 'PYKNOCYTOSIS'. The pyknocyte is a less well-defined entity. Tuffy, Brown, and Zuelzer (1959) gave the name to distorted and contracted erythrocytes which they found in the blood of many apparently normal full-term infants. These cells were more numerous in normal premature infants but, at most, comprised only 5-6% of all red cells whereas up to 50% of red cells had this appearance in 11 newborn babies with an unexplained haemolytic anaemia. This anaemia was severe enough in some cases to require transfusion, but then resolved spontaneously with disappearance of the abnormal cells. Tuffy *et al.* considered these

cells to be morphologically identical to burr cells but proposed the term pyknocyte because the density of the cells was regarded as a fundamental feature and resembled that of spherocytes. The cells were also regarded as identical in appearance to acanthocytes. Lovric (1960), however, in reporting the occurrence of pyknocytosis in twins with haemolytic anaemia compared the appearances of the red cells with those from a case of acanthocytosis. He found that in pyknocytosis abnormal cells were fewer in number, but that almost all had the appearance of 'contracted spherocytes with pseudopodia', whereas in acanthocytosis out of 70% irregular cells only a proportion had this appearance.

To summarize, there are three conditions in which contracted, deeply-staining, highly irregular, spiny red cells occur, but consideration of the over-all appearance of the blood film will enable the correct clinical correlation to be made. In acanthocytosis the cells will comprise one-third to one-half of all cells and will be accompanied by many showing normal crenation. In the burr cell phenomenon, only a small proportion of abnormal cells will have this appearance, the remainder will show a wide variety of irregular shapes among which triangular, crescentic, and helmet-shaped cells will be prominent. In pyknocytosis it has been stated that one-third to one-half of cells will resemble the acanthocyte but there will be few if any other abnormal cell types.

Acanthocytes retain their abnormal shape when incubated in normal serum. Conversely the serum of patients with this disorder does not convert normal cells into acanthocytes. The abnormality would, therefore, appear to be inherent in the erythrocyte rather than in the serum. It has been shown that acanthocytes can be converted into the shape of normal erythrocytes by exposure *in vitro* to a non-ionic detergent (Switzer and Eder, 1962) and *in vivo* by administration of cotton-seed oil emulsion intravenously (Di George, Mabry, and Auerbach, 1961).

Other constant haematological features in acanthocytosis are absence of rouleaux formation and a low erythrocyte sedimentation rate. Our case had an erythrocyte sedimentation rate of 3 mm. (Westergren) at the height of a respiratory infection.

Anaemia is not a constant feature in this disorder. In three patients the half life of the erythrocytes has been shown to be shortened by the radioactive chromium method (Druez, 1959; Mier *et al.*, 1960; Ways *et al.*, 1963). The anaemia in our patient appeared to respond to intramuscular iron and folic acid and presumably was nutritional in origin.

Unusual Features. Our patient showed several

unexplained features which have not been noted in previous cases, these features being recurrent pulmonary infections, impaired renal function, and aminoaciduria. The recurrent pulmonary infections suggested the possibility of mucoviscidosis but the sweat sodium and chloride concentrations were normal and the stools showed proteolytic activity to a titre of 1 : 300.

The nature of the renal lesions causing impaired renal function and aminoaciduria was not ascertained in our patient. We do not know whether the impaired renal function, renal aminoaciduria, and recurrent pulmonary infections were related to the Bassen-Kornzweig syndrome or not. As these features do not seem to have been present in the 12 previously reported cases with this disease, it is possible that they are unrelated.

A final puzzling feature in our patient was the development of cardiac failure with pulmonary oedema during an episode of bronchitis and asthma. This complication contributed to his death and is unexplained. Schwartz, Rowland, Eder, Marks, Osserman, Anderson, and Hirschberg (1961) in their review of Bassen and Kornzweig's second case mention the presence of cardiomyopathy. Their diagnosis of cardiomyopathy was based on the presence of a loud systolic murmur. Whether cardiomyopathy can occur as a complication of the Bassen-Kornzweig syndrome will only become apparent by study of a greater number of cases. Cardiomyopathy has been reported in other mal-absorptive states (McGiven, 1962).

Aetiology. Di George *et al.* (1961) have suggested that the clinical and biochemical abnormalities are all secondary to a genetically determined inability to absorb and transport lipids. Salt *et al.* considered that the primary gene defect was inability to form the β -lipoprotein molecule. They found subnormal β -lipoprotein levels in both parents and the paternal grandfather of their patient. However, in the family of the patient we are describing and in several other families reported serum β -lipoprotein levels have been normal (Wolff and Bauman, 1961; Lamy *et al.*, 1963; Ways *et al.*, 1963; Schwartz *et al.*, 1963). These investigations do not confirm the presence of biochemical abnormality in the presumed heterozygous state, but the familial occurrence (Kornzweig and Bassen, 1957) and frequency of parental consanguinity remain as evidence for the disease being due to an autosomal recessive gene. Salt *et al.* also favoured Singer's view that the ocular and nervous disturbances were genetically determined, and speculated that a factor essential for the production of β -lipoprotein was also essential for the

normal structure and function of nerve cells. However, they did not dismiss the possibility that these disturbances result from abnormal fat absorption and transport.

Summary

A 17-month-old Maori child with Bassen-Kornzweig syndrome is described. As well as showing acanthocytosis, α - β -lipoproteinaemia, hypocholesterolaemia, and steatorrhea, he had incipient neuropathy.

The child also had impairment of renal function, aminoaciduria, and suffered from recurrent pulmonary infections. None of these features appear to have been described in other patients with this disease and therefore they may be unrelated to the primary disorder.

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