

Bassen F A & Kornzweig A L. Malformation of the erythrocytes in a case of atypical retinitis pigmentosa. *Blood* 5:381-7, 1950.
[Consultation Service, Mount Sinai Hospital, New York, NY]

A very rare hereditary disease, first described in 1950, continues to be of interest despite its infrequent occurrence. This is due to the discovery in 1960 of an inborn error of metabolism, α -beta-lipoproteinemia, and the questions this discovery raised as to its relationship to the complex clinical and laboratory findings. [The SC¹® indicates that this paper has been cited in over 205 publications since 1955.]

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November 16, 1985

The circumstances regarding our studies did not at the onset seem very unusual. The patient was an 18-year-old Jewish girl referred to the Consultation Service at the Mt. Sinai Hospital for diagnostic studies. To state matters briefly, she had a clinical picture like that seen in Friedreich's ataxia and retinitis pigmentosa with considerable loss of vision. There was also a history of protracted steatorrhea in early childhood. It was only when it was discovered that she had most unusually and bizarrely shaped red blood cells that we became intensely interested. Family blood studies revealed the same type of red cells in a nine-year-old brother, who was at that time relatively asymptomatic. He also had early pigmentary retinal changes. This convinced us that we were dealing with a hereditary disease not previously described.

Two years after publication, with no further cases reported and very few requests for reprints, we began to suspect it wasn't creating much interest and was exceedingly rare. We were somewhat relieved when, in

1952, Singer¹ and his associates reported a remarkably similar case and in addition conferred the term acanthocytes (later changed to acanthocytes²) on the bizarrely shaped red cells.

A few more cases were reported, but it was not until 1960 that Salt³ and his associates reported a new and exciting finding. Their patient, an 18-month-old child, had a history of protracted steatorrhea, an inordinately low blood cholesterol (22 mgm %), and acanthocytosis. In-depth studies on fat metabolism were undertaken. The blood lipoprotein determination showed a complete absence of beta lipoprotein. The investigators considered this finding to be an integral part of the complex disease process and suggested that the term α -beta-lipoproteinemia be used to describe the syndrome. This term has since become part of the medical literature.

Whether the absence of the beta lipoprotein alone could be responsible for the entire variegated clinical picture is a matter of conjecture. In any event, its absence provided investigators with something tangible to speculate about and work with. I assume that the frequent citation of our work is because we described the first case of this rare disease. The continued interest in it is undoubtedly due to the discovery of the inborn metabolic error, which, although known, is not clearly understood. Our role was important in calling attention to the existence of this rare and unusual hereditary disease, but the continued interest has obviously been engendered by those who came after. Despite its infrequent occurrence, a fairly recent review (1983) of the literature⁴ revealed that over 50 cases have now been reported. For those who might be interested in a more detailed account of this syndrome, a very satisfactory review may be found in the textbook, *The Metabolic Basis of Inherited Disease*.⁴

1. Singer K, Fisher B & Perlstein M A. Acanthocytosis: a genetic erythrocytic malformation. *Blood* 7:577-91, 1952. (Cited 85 times since 1955.)
2. Druet G. Un nouveau cas d'acanthocytose: dysmorphie érythrocytaire congénitale avec rétinite, troubles nerveux et stigmates dégénératifs. *Rev. Hematol.* 14:3-11, 1959.
3. Salt H B, Wolff O H, Lloyd J K, Fosbrooke A S, Cameron A H & Hubble D V. On having no beta-lipoprotein: a syndrome comprising α -beta-lipoproteinemia, acanthocytosis, and steatorrhea. *Lancet* 2:325-9, 1960. (Cited 220 times.)
4. Herbert P N, Assmann G, Gotto A M, Jr, & Fredrickson D S. Familial lipoprotein deficiency: α betalipoproteinemia, hypobetalipoproteinemia, and Tangier disease. (Stanbury J B, Wyngaarden J B, Fredrickson D S, Goldstein J L & Brown M S, eds.) *The metabolic basis of inherited disease*. New York: McGraw-Hill, 1983, p. 589-621.