The Acanthocyte-Echinocyte Differential. The Example of Chorea-Acanthocytosis

A. Foglia

via Bosia 4, CH-6900 Lugano-Paradiso

dralberto.foglia@bluewin.ch
Keywords: Acanthocytes, Echinocytes, Chorea-acanthocytosis (ChAc)

Chorea-acanthocytosis (ChAc) is a progressive untreatable neurodegenerative disorder correlated with a deformation of the red blood cells (RBCs) called acanthocytosis (from acantho- "thorn", "spur cell"). ChAc is part of a clinical syndrome group called Neuroacanthocytosis syndromes (NA), first described in 1960 as "Levine-Critchley syndrome" [1]. Neuroanatomical changes are present in form of extensive neuronal loss and gliosis of the caudatum, the corpus striatum and the pallidum and peripheral axonal neuropathy [4,5]. The concomitant neuronal degeneration and erythrocyte membrane abnormality may have a common proteic source (6), distinct from the lipidic source of acanthocytes of other aetiologies (M. Anderson, abetalipoproteinaemia, hypobetalipoproteinemia, alcoholic liver cirrhosis, anorexia nervosa) [7].

Microscope images of peripheral blood smears, especially scanning electron microscopic ones, are reported as useful tools in investigating and assuring diagnosis of NA [4,8,9]. Our scanning electron microscopic investigation makes it possible to objectify the morphology of RBC in ChAc in detail, in fact the abnormality of the acanthocytic-transformed erythrocytes is very pronounced, sometimes grotesque. This confirms the assumption that acanthocytes are a distinct structural (and functional) entity compared to echinocytes (from echino- "porcupine", "burr cell")[10], a differential which sometimes has been confused.

Methods: EDTA-blood sample from an advanced Chorea-acanthocytosis (ChAc) clinical case with severe choreo-athetoid movement disorders, orofacial dyskinesia and dementia is fixed in 2,5% glutaraldehyd and stored at room temperature for 24h in Sörensen solution. After three washing procedure (centrifugation in bidest. H2O), the solution is dehydrated in increasing concentrations of acetone (20, 40, 60, 80, 95 and 100%; 10 min each), placed on Poly-l-lysine coated 6 mm coverslips and air dried for 2h. Platin coating was performed with a Balzers SCD 004 sputter coater and visualized with a Scanning Electron Microscope Jeol JSM 840, with 15,0 kV accelerating voltage, magnification 1'400x, 6,000x 35,000x (Fig. 1), 13,000 (Fig. 2), 7,500x (Fig. 3). The light-microscopic images (Fig. 1,2) were recorded with a Zeiss Axiovert 200 M, camera Sony DSC-S85, standard preparation and stain from EDTA blood from the same patient. Control blood from an hepatocellular carcinoma suffering patient.

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I thank Dr.Valerio Saglini and Dr. Gianantonio Rosti for reviewing the present paper. I thank the patient and her family for providing me the blood to investigate.

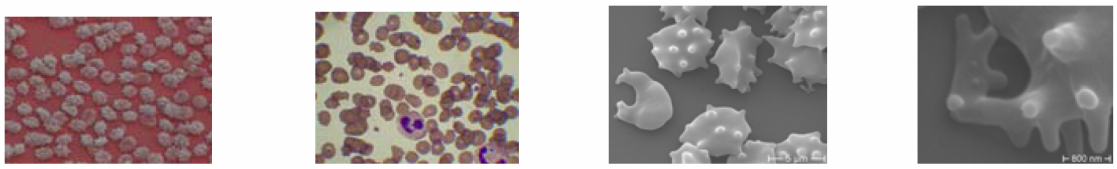

Figure 1. Chorea-acanthocytosis: acanthocytic and echinocytic deformation of RBCs. Peripheral blood smear: acanthocytes > 5-10%. Schizocyte, acanthocytes and echinocytes in concomitance. Gross deformation of RBC membrane in acanthocyte.

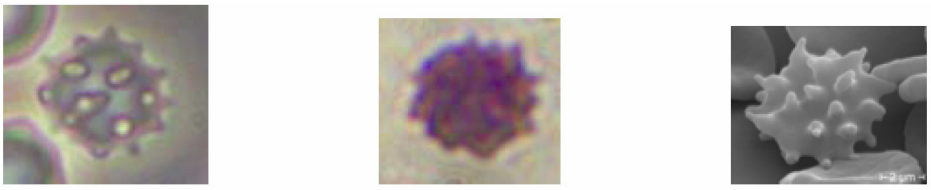

Figure 2. Echinocyte in normal control: numerous spiculae regularly distributed in living specimen (DIC 945x), standard staining (BF 1000x) and Scanning Electron Microscope.

Figure 3. Acanthocyte in ChAc: fewer irregularly distributed spiculae, in living specimen (DIC 945x), standard staining (BF 1000x) and Scanning Electron Microscope.

DOI: 10.3217/978-3-85125-062-6-269, © Verlag der TU Graz 2009