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LETTER TO THE EDITOR

Remarks on the article Genetics and immunopathology of chronic granulomatous disease by Marie José Stasia and Xing Jun Li

Joachim Roesler

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The article by Marie José Stasia and Xing Jun Li, published in Seminars in Immunopathology, 2008 May 29., Epub ahead of print, PMID: 18509647, is excellent in most parts.

However, some views of the authors in the chapter about "CGD diagnostic tests" are not shared by many clinicians and researchers in the field. I wonder why the authors do not cite any reference in the whole chapter. This gives the impression that the authors just describe their own practice without considering the experience of others.

Colleagues of mine and I have encountered several missed and false diagnoses of chronic granulomatous disease (CGD) by the nitroblue tetrazolium (NBT) slide test (which the authors recommend as sufficient for testing) especially when shipped blood was used. Neutrophils with residual NADPH activity were considered normal, and in one instance, even a patient without such residual activity was missed (possibly because of an electron leak of mitochondria in damaged cells). On the other hand, damaged cells sometimes led to a false assumption of CGD.

In contrast, the flow cytometric dihydrorhodamine (DHR) test worked well in the hands of experienced lab technicians—not only with freshly drawn but also with shipped blood. When cells are damaged by shipping, often a subpopulation of cells still gives a normal signal. At least, we have never encountered a false normal result with the DHR assay, but false pathologic results do occur. These considerations and respective references are summarized in Mauch et al. (2007) Clin Chem 53(5):890–896 (May).

The truth is that no single test for CGD whatsoever is always reliable under all circumstances. Consequently, all suspicious results should be re-tested using a second blood sample from the patient and a second method. This should be emphasized, because the diagnosis of CGD is essential for the life of the patient.

Furthermore, I would like to add a remark to the chapter about management of CGD: Careful regular surveillance (e.g. pulmonary function tests, abdominal ultrasound) of patients is essential for their prognosis. Therapy is most effective if applied before they suffer from symptoms (e.g. abscesses in the liver can go unnoticed for a long time and can be huge by the time the patient gets a fever or feels pain; slow development of fibrosis of the lungs can also go unnoticed for long). Further examples are published by Roesler et al. (2005) J Eval Clin Pract 11(6):513–521 (Dec).

Nevertheless, I would like to emphasize that most parts of the article such as genetics and physiology of CGD provide excellent overviews.

J. Roesler (✉)
Department of Pediatrics, University Clinic Dresden,
01307 Dresden, Germany
e-mail: roeslerj@rcs.urz.tu-dresden.de