Asymptomatic elevation of creatine kinase (CK) was first reported by Rowland, who proposed to call the condition “high serum enzymes, cause undetermined.” In 1980, Rowland et al. coined the term “idiopathic hyperCKemia” to describe patients with serum creatine kinase levels that were consistently higher than normal despite lack of symptomatic weakness or other abnormality on neurological examination, electromyography, or muscle biopsy. Additional criteria for the diagnosis of idiopathic hyperCKemia were proposed in 1988 by Brewster and de Visser. These included absence of signs of hyperthyroidism, family history of neuromuscular disease, intake of medications that produce hyperCKemia, and intramuscular injections. No critical levels of serum creatine kinase activity were set to define the condition, and there was no follow-up of patients to determine their outcome.

With increasing concern about malignant hyperthermia and with the inclusion of creatine kinase determination in the automated blood chemistry profile performed upon hospital admission or as part of health screening, the number of patients with unexplained rise in creatine kinase has increased.

A literature search for patients with idiopathic hyperCKemia yielded a total of 55 patients, of whom only seven are children. The number of literature reports on idiopathic hyperCKemia has drastically diminished since the discovery of dystrophin and its gene and the subsequent development of knowledge regarding the variable presentations of a dystrophinopathy. In our institution, four children with the diagnosis of idiopathic hyperCKemia were encountered over a period of 10 years.

Reports in the literature on idiopathic hyperCKemia reflect a male preponderance. Of reported patients, 78% of cases of all ages and 71% of childhood cases are male. Reported serum creatine kinase values vary from < 5-fold above the normal value up to an increase of 170-fold over normal. The majority have serum creatine kinase values between 5- and 10-fold above normal. No relationship between magnitude of serum creatine kinase elevation and age has been reported. None of the reported patients had weakness on initial examination, and about one third reported muscle-related symptoms, including myalgias, fatigue, cramps, and stiffness. Electromyographic (EMG) abnormalities are described in 41% of reported patients and in 56% of childhood cases. The commonest EMG abnormalities are a myopathic pattern, short duration potentials alone or with positive sharp waves, and fibrillations. Abnormalities in muscle biopsy specimens are reported in 56% of patients. Variations in fiber size, necrotic and regenerating fibers, and inflammatory myopathy are the most frequently reported findings. The family history of reported patients is usually normal with occasional reports of prominent calves.

The duration of follow-up in reported cases varies from 1 to 12 years; the majority were followed for 4 years. Clinical and/or histopathological evidence of a neuromuscular disorder developed between 1 and 7 years after detection of hyperCKemia in one third of the cases. These include distal myopathy, myoadenylate deaminase deficiency, polymyositis, mitochondrial myopathy, sarcoid myopathy, McArdle’s disease, central core disease, multicore disease, inclusion body myopathy, and a carrier of Duchenne muscular dystrophy. A muscle biopsy was needed for the diagnosis in the majority of cases. Thus, without a muscle biopsy, the diagnosis of idiopathic hyperCKemia remains uncertain. There are no clues at the onset of hyperCKemia to help predict the outcome.

The subsequent development of neuromuscular disorders in patients who present with idiopathic hyperCKemia calls for vigilant follow-up especially in those patients with electromyographic and/or muscle biopsy abnormalities. Furthermore, the detection of carrier state for dystrophinopathy in female patients with idiopathic hyperCKemia in Tachi’s report, and in one of our patients, suggests that tests for carrier state of a dystrophinopathy be done on all patients with idiopathic hyperCKemia, and that absence of dystrophinopathy be added to the criteria proposed by Rowland and Brewster and de Visser for the diagnosis of idiopathic hyperCKemia.
References


The 21st Annual Carrell-Krusen Symposium

A Call for Abstracts

Abstract Deadline: Nov. 23, 1998

The 21st Annual Carrell-Krusen Symposium, to be held Feb. 25-26, 1999, at Texas Scottish Rite Hospital for Children in Dallas, focuses on the treatment of neuromuscular disease and changes in current clinical practice. Guest lecturer will be Robert G. Miller, M.D., Director of the Neuromuscular Research Center at the California Pacific Medical Center, San Francisco, California.

Abstracts for submission should be prepared on a single sheet of plain white paper. Place the complete title, in upper case, on the first line followed by the name and city location of each author underneath. Limit abstract titles to 65 characters. Skip one line and indent three spaces to begin abstract text. Abstracts must be double-spaced and one paragraph in length, with a maximum of 300 words. At the bottom of the page, give name, academic and position titles, mailing address and phone and fax numbers of the presenting author. Mail original, 10 copies and a computer disk labeled with the software package and file format to: Susan T. Iannaccone, M.D., Department of Neurology, Texas Scottish Rite Hospital for Children, 2222 Welborn Street, Dallas, TX 75219, or call 214/559-7830 for information. Accepted abstracts will be published in the Journal of Child Neurology and must not have been presented or published before the meeting.

A cover letter included with the abstract and signed by all authors must contain the following text: "The author(s) has/have read and agree with the content of this abstract submitted for the 1999 Carrell-Krusen Symposium and warrant(s) the material is (1) original work of the author(s), (2) does not violate my copyright proprietary or personal rights of others, (3) is factually accurate and contains no matter libelous or otherwise unlawful, (4) has not been, nor will be, published or presented elsewhere prior to the 1999 Carrell-Krusen Symposium, and (5) hereby transfers, assigns or otherwise conveys all copyright ownership of this abstract to the Journal of Child Neurology and Decker Periodicals. In addition, the author(s) agree(s) to acknowledge all commercial support for options, royalties, consulting fees and honoraria for speaking material support and other financial arrangement(s) with the manufacturer(s) of any commercial product or service relating to the abstract by any author has been described fully in this cover letter."

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