Gaucher disease in a 53-year-old Iranian man

Sir,

Gaucher disease (GD) is a rare genetic disorder. Glucocerebrosidase deficiency affects metabolism of cellular glycolipids, thus glucocerebroside deposits in the lysosomes of cells. GD is more common in the Ashkenazi Jewish population.[1]

Non-neuronopathic form (type-1, GD1) includes more than 90% of cases.[2] Acute neuronopathic form (type-2, GD2) includes 1% of cases and causes to death during infancy or the 1st years of life.[3]

Subacute or chronic neuronopathic form (GD3) includes 5% of cases and has a slower and later onset than GD2 and leads to death during childhood or early adulthood.[3]

A 53-year-old man was admitted with pancytopenia. He had history of easy bruising and prolonged bleeding following dental extraction for a long period. He had no fever, night sweat, and weight loss. Physical examination revealed massive hepatosplenomegaly. Laboratory findings were included: WBC = 2,800/μl, Hb = 8.5 g/dl, and PLT = 15,000/μl. Erythrocyte sedimentation rate (ESR), renal and liver function tests were normal. Abdominal sonography showed a huge nodular splenomegaly. Bone marrow aspiration showed a hypercellular polymorph marrow with increased megakaryopoiesis and an increased in number of macrophages with a crumpled tissue paper appearance and displaced nuclei (Gaucher cells) [Figure 1]. Bone marrow biopsy showed sheets of Gaucher cells [Figure 2]. The diagnosis of GD was made. Patient's blood sample was sent to Austria for enzyme assay.

GD1 has asymptomatic or symptomatic variants. Symptomatic form present during childhood or the late adulthood but seldom may present in elderly.[4,5]

In this 53-year-old Iranian Muslim with GD1, all of his six siblings were asymptomatic in ages between 60 years and 80 years. Although he had prolonged bleeding and easy bruising for a long period, he did not take any medical attention. GD1 can occur in higher ages without positive family history. We should consider GD1 in differential diagnosis of pancytopenia and organomegaly in old age patients even in the absence of positive family history for GD.

ACKNOWLEDGMENT

We express our appreciation to Drs. Abbas Saffarifard and Aida Gheibi, residents of the Pathology Department in Urmia University of Medical Sciences who helped us in taking photo from pathology slides of this patient.
Chronic disease: Working together

Sir,

I enjoyed reading the editorials on Dr. Rita Levi-Montalcini as well as on patient centered care,[1,2] and I would like to share a story about an admirable Estonian woman Maire-Liis Hääl. She is not as famous as Dr. Rita Levi-Montalcini. She is an ordinary woman, but ordinary persons can also achieve extraordinary feats.

One autumn several years ago, a woman introduced herself as chairwoman of Tallinn Rheumatism Association and explained her wish to study the problems and needs of people with rheumatism. In order to collect and analyze data in a scientific manner, she needed the help of specialists. She talked energetically and passionately, and since her inner strength and wish to act were so enormous, I did not need a lot of convincing to take part in the study.

Maire-Liis Hääl formed a maximally effective research group. In addition to herself, who suffered from rheumatism and knew all the problems related to this disease, she included one of the best rheumatism doctors, a professor emeritus of University of Tartu, and me, a sociologist. My task was to carry out the data analysis.

This multi-disciplinary research team facilitated a holistic study of the life of rheumatism patients. We studied not only the problems related to the illness itself, but also their life in general, including their coping skills, strengths and weaknesses. Data collection took place in cooperation with Rheumatism Associations, rheumatologists and general physicians all over Estonia. 1450 questionnaires were distributed, out of which 808 were filled and returned. As a result of this cooperation we published a book, coping with life and availability of treatment for people with rheumatism in Estonia.[3]

Maire-Liis Hääl was able to see the situation on a wider scale – apart from the biological aspect of disease, she calculated its economic impact as well. In healthy people, 90% of all health-related expenditure was on health promotion like nutraceuticals, rehabilitation, and hobby sports (massage, pool, tennis, gym). In people with illness, expenses on rehabilitation were very small (7%), due to which 64% said the possibility of achieving rehabilitation was poor.[3]

What did we learn from this cooperation and why am I writing to you?

First, reviewers said the composition of the research team was good and optimal. Because what one lacked, the other contributed. So it was research and practice all together, combining clinical care, sociology and patient perspectives. The team was led by a person with rheumatism, not by a clinician. Second, we studied more thoroughly people who suffered from one type of disease. This focused approach helped us achieve more tangible results. Furthermore – we translated the shortened version of the questionnaire into Finnish, as the Vantaa Rheumatism Union and also Nordic Rheumatism Unions thought that it would be important for them to carry out a similar study. Thirdly, it should be emphasised that our work had major practical implications. The society is unaware of what people with rheumatism and their families have to suffer in order to cope with everyday life, their expenses, and changes in quality of life. However, people who are ill sense the negative attitude of society, due to which it is necessary to change the opinions of the public, patients and specialists about people suffering from rheumatism. We showed what mistakes people with rheumatism had made themselves (go to the doctor too late, stop taking medicine...