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Commentary: The Natural History of Gaucher Disease

Almost a decade ago I saw in consultation a 23-year old woman with very mild Gaucher disease. She had been told by her physician that she had three or four years to live. "I have seen cases of Gaucher disease before," he explained. Fortunately, such gross misperceptions on the part of the medical profession are uncommon. Yet, apart from our studies of the natural history of the disease (1-3), there was little information in the literature that provided guidance as to what was likely to happen in the future to patients with Gaucher disease. Such knowledge is particularly important if one is to apply treatment modalities that are very expensive, such as enzyme replacement, or hazardous such as stem cell transplantation. We concluded from our observations that there was very little progression in most adults with Gaucher disease; progression occurs almost entirely in the very young. In spite of this, "prophylactic" therapy of adult patients with stable disease is often undertaken at great and unjustified cost.

The study of Ida and colleagues (4), which carefully documents the clinical manifestations in 35 Japanese type 1 Gaucher disease, is an especially important contribution to our knowledge of the natural history of Gaucher disease. Among the Japanese the disease is particularly severe and, as Ida et al. rightly point out, this is almost surely due to the fact that the 1226G(N370S) mutation is not found in Asian patients. It is this functionally mild mutation that predominates in Ashkenazi Jews and also accounts for one-third to two-thirds (5,6) of the mutations in non-Jewish European patients with Gaucher disease.

Because of the disease severity, observations on most of the patients in Japan were started at a very young age. There were exceptions, however. In the case of four patients, observations were started when the patient was over 25 years of age. Calculating the yearly rate of change of the severity score index (SSI) provides the striking results shown in Figure 1. It is such data, not vague impressions often inappropriately translated without justification from one age to another, that need to be taken into account in making treatment decisions.

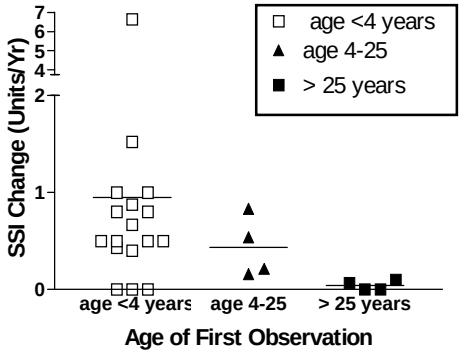

Figure 1. The average annual changes in the severity score index in the data reported by Ida et al. (4).

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