THE ROCKEFELLER UNIVERSITY
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July 9, 1995

Rabbi Josef Ekstein
Executive Director
Dor Yeshorim
Committee for Prevention of Jewish Genetic Diseases
160 Wilson Street, 1-B
Brooklyn, NY 11211

Dear Rabbi Ekstein:

I strongly support Dor Yeshorim's decision to expand its testing program to include Fanconi anemia. During our pilot study of 3,104 individuals with no family history of FA, we identified 35 carriers, for a carrier frequency of 1 in 89. Among these carriers we identified one at risk couple, saving them from the potential tragedy of having children with this terrible disease.

Fanconi anemia is an autosomal recessive syndrome characterized by multiple birth defects, bone marrow failure and predisposition to cancer, especially acute myelogenous leukemia. Birth defects may include absent thumbs and radii, other skeletal malformations, as well as abnormalities of the kidneys, heart, gastointestinal tract, genitalia and central nervous system. Children usually have low birth weight, and grow slowly. Onset of bone marrow failure typically occurs around the age of two years. Complications of bone marrow failure, or leukemia are the usual causes of death in children with Fanconi anemia.

I have seen the affects of this devastating disease in my work with families in the orthodox community. Families with several affected children have had to cope with the death of one child after another. Until recently, couples had no way of knowing if they were at risk of having a child with Fanconi anemia before the birth of their first affected child. As in the case of Tay-Sachs, carriers themselves are completely healthy. The discovery that a single mutation in a gene for Fanconi anemia is the usual cause of this disease in the Jewish population has led to the development of a simple carrier test. With the availability of such a simple test, we recommend that the orthodox community add Fanconi anemia to their genetic screening programs.

Sincerely yours,

Arleen D. Auerbach

Arleen D. Auerbach, PhD
Associate Professor
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