

B'SHALOM COLUMN
American Association on Mental Retardation
Religion and Spirituality Division

Jewish Genetic Diseases

Recently, I was asked to participate in a conference on genetics to speak about the genetic diseases specific to Jewish individuals. There are at least seven genetic diseases, with research being done on several more. They include Tay-Sachs, Gaucher, Neimann-Pick, Canavan, dystonia, familial disautonomia, and mucopolipidosis type 4. Autism, bi-polar and schizophrenia are currently being studied for the possible Jewish connection; breast cancer genes (BRCA 1 and BRCA 2) have already been identified as being prominent in Jewish women.

For centuries, Jews lived separately from the non-Jews in whatever country in which they resided, either by choice or by force. Ghetto living bred a limited number of potential marriage partners, and that is likely to have contributed heavily to the genetic problems of today. The expectation to marry within the Jewish religion, long after ghetto living ceased, continues to nurture an undiluted gene pool. The ramifications can be devastating.

Genetic studies all focus on Ashkenazic Jews, those of Eastern European and Russian backgrounds. Sephardic Jews, those of Mediterranean, Spanish and Portuguese background, are not mentioned in the studies. Tay-Sachs, an enzyme deficiency which causes a degenerative and deadly condition in infants, was one of the earliest identified Jewish genetic disorders. *One in every 25 Ashkenazic Jews is a carrier.* Gaucher disease, another enzyme deficiency, affects the liver, spleen and bone marrow, causing increased bleeding and anemia, and *one in every 13 Ashkenazic Jews is a carrier.*

Knowing these statistics creates troubling questions in the Jewish community.

For decades, rabbis have suggested genetic testing for at least Tay-Sachs disease when they provide pre-marital counseling. Should they urge more extensive testing? Should marriages of "carriers" be discouraged, or should the couple be advised not to have children? Other questions arise regarding sharing this knowledge in negotiating the "arranged marriages" which still occur in the ultra-Orthodox Jewish communities.

As the parent of a 22 year old daughter, I have always assumed that she would marry a nice Jewish boy and raise little Jewish grandchildren for me to spoil. Now, knowing that our families' exclusively Ashkenazic background puts her at risk, knowing that her brother's autism may soon join the list of Jewish genetic disorders, I am forced to reevaluate my assumptions.