NATIONAL HEMOPHILIA FOUNDATION
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MASAC Document #214
(Replaces Document #96)

The following recommendation was approved by the Medical and Scientific Advisory Council (MASAC) on November 10, 2012, and adopted by the NHF Board of Directors on November 11, 2012.

MASAC RECOMMENDATIONS ON THE NHF GENOTYPING PROJECT FOR PERSONS WITH HEMOPHILIA

At this Annual Meeting it was announced that NHF has formed a partnership to provide genotyping for the hemophilia community. *My Life, Our Future, Genotyping for Progress in Hemophilia* is a 4-way partnership between the National Hemophilia Foundation (NHF), the Puget Sound Blood Center (PSBC), American Thrombosis Hemostasis Network (ATHN), and Biogen Idec. NHF will educate and publicize the Genotyping Project to individuals with hemophilia; PSBC will perform the DNA analysis and send results to the ordering HTC; and with patients' permission, ATHN will put the patients' de-identified genotypes into the ATHN dataset. Biogen Idec will provide funding for the project.

Since the genes for factors VIII and IX were identified and sequenced in the 1980s, numerous gene defects have been identified in persons with hemophilia A and B. This information has led to increased understanding of the molecular biology of these genes and has established new correlations between a person's genotype and phenotype. High through-put technology and strategies for more efficient genotyping have reduced the costs of doing genotyping significantly.

In 1998, MASAC recommended that NHF identify public and private funding sources that could facilitate widespread genotyping efforts in the hemophilia community. In 2003, this concept was endorsed but not funded by Congress. Another event that made a community-wide genotyping project feasible was the passage in 2008 of the Genetic Information Nondiscrimination Act (GINA). This act guarantees that genetic information cannot be used to discriminate against an individual.

With this endorsement from Congress and the protections in GINA, NHF began to look for private partners to support a community-wide genotyping initiative; this initiative culminated in *My Life, Our Future*.

My Life, Our Future will provide genotype testing for individuals with hemophilia and later for potential carriers. This information will allow individuals and healthcare providers to predict bleeding severity, determine inhibitor risk, identify carriers, help with family planning, and lead towards improved, individualized treatments. Also, with the emergence of experimental gene therapies, genotyping is important in predicting those individuals who might be potential candidates and in selecting the most appropriate form of gene therapy for each individual.

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When individuals come to their HTC for their Genotyping Project blood draw, they will be asked if they will agree to have their mutation added to the ATHN dataset. All data added to the ATHN dataset is de-identified, that is, all information that could link the mutation to a specific individual is removed before adding the data in an anonymous way to the dataset. The dataset is maintained in a confidential manner with strict limits on who has access to the data. Researchers, including academic institutions and pharma, who wish to use the data must apply to the ATHN Research Project Review Committee for permission.

An important aspect of genotyping is genetic counseling. HTCs who agree to participate in the Genotyping Project will have genetic counselors available to counsel individuals and families pre- and post-genotype testing to ensure that they understand the implications of the test results.

Therefore, MASAC makes the following recommendations:

1. MASAC recommends that all individuals with hemophilia and their affected family members participate in the Genotyping Project.
2. MASAC recommends that HTCs participate in the Genotyping Project by informing patients and families of its availability and by drawing samples for genotype analysis at PSBC.
3. MASAC further recommends that individuals and family members strongly consider agreeing to have their de-identified mutation results added to the ATHN dataset in order to further research into an understanding of the relationships between genotypes and clinical phenotypes.
4. MASAC urges the maintenance of strict guidelines for confidentiality of and access to the dataset and repository samples.