



## Mid-Atlantic Region III Hemophilia Genetics Information Toolkit

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The following is a list of articles, brochures and other resources that may be helpful to you as clinical HTC staff in communicating information regarding the genetics of hemophilia to your patients. You should review each of these prior to sharing them with your patients to ensure that he/she is at the right reading level and at the right stage in life for the information. Not all of these resources are appropriate for each patient. You should tailor the resources that are shared with a patient and family over the course of their life span to address their concerns as they encounter different challenges.

Each resource has a brief description, guidance on whom it may be appropriate to share with, and if the resource has any limitations. There should also be a link or an email address to obtain a copy of the resource for your patients. This list will be updated on an annual basis to help ensure that the most up to date information is available for you and your patients.

### I. Resources for Carriers

1. **All About Carriers**, by the Canadian Hemophilia Society
  - a. Download a copy at: <http://www.hemophilia.ca/en/women/educational-resources/> under Carriers of Hemophilia A and B section
  - b. Guidance: This document provides a thorough overview of all aspects related to being a carrier or potential carrier of hemophilia.
2. **Carriers and Women with Hemophilia**, by World Federation of Hemophilia
  - a. Download a copy at: <http://www.wfh.org/en/abd/carriers/carriers-resources-en>
  - b. Guidance: This document provides an overview of symptomatic and asymptomatic carriers of hemophilia.
3. **Symptomatic Carriers of Hemophilia**, by World Federation of Hemophilia
  - a. Download a copy at: <http://www.wfh.org/en/abd/carriers/carriers-resources-en>
  - b. Guidance: This resource describes common bleeding problems in symptomatic carriers and strategies to prevent and treat them.
4. **Carrier Fact Sheet**, Hemophilia Center of Western Pennsylvania
  - a. Available by request from Michelle Alabek at [malabek@itxm.org](mailto:malabek@itxm.org)
  - b. Guidance: This fact sheet can help to facilitate family communication about information important for carriers and potential carriers of hemophilia.
5. **At-Risk Relative Identification Card**, Hemophilia Center of Western Pennsylvania
  - a. Available by request from Michelle Alabek at [malabek@itxm.org](mailto:malabek@itxm.org)
  - b. Guidance: This card can be filled out and given to patients to outline for them which of their relatives may benefit from being evaluated for a possible bleeding disorder.

6. **Template Family Letters**, University of Virginia HTC and the Children’s Hospital of Philadelphia (CHOP) HTC
  - a. Available from the Core Center by contacting Danielle Deery at deeryd@email.chop.edu
  - b. Guidance: These template letters can be customized and provided to patients for them to share with their family members who may benefit from testing or further genetic education.
7. **“Knowledge is Power for Symptomatic Carriers,”** an article in NHF’s Hemaware magazine
  - a. Download a copy at: <http://www.hemaware.org/story/knowledge-power-symptomatic-carriers>
  - b. Guidance: This article may be helpful for women who are symptomatic carriers or who have recently had hemophilia diagnosed in their families. It can help them understand how the diagnosis may affect them.
8. **“Testing Carrier Factor levels in Women,”** an article in NHF’s Hemaware magazine
  - a. Download a copy at: <http://www.hemaware.org/story/testing-carrier-factor-levels-women>
  - b. Guidance: This article could be helpful to some patients who are interested in getting genetic testing to determine whether or not they are a carrier.
  - c. Limitation: The information could be considered somewhat outdated, since it does not clearly convey that factor level alone is not definitive of carrier status.
9. **“Hemophilia Carrier Testing,”** a page on the Steps for Living website
  - a. Download a copy at: <http://www.stepsforliving.hemophilia.org/basics-of-bleeding-disorders/genetics-of-bleeding-disorders/hemophilia-carrier-testing>
  - b. Guidance: This page provides information regarding who may be a carrier, testing for carrier status and when to have testing performed on suspected carriers.
10. **“Hemophilia: Information for Women,”** a page on the CDC website for women
  - a. Download a copy at: <http://www.cdc.gov/ncbddd/hemophilia/women.html>
  - b. Guidance: This page provides information on carriers, pregnancy and childbirth and some care considerations for newborns.
11. **“Family Planning for Carriers,”** an article from NHF’s HemAware magazine
  - a. Download a copy at: <http://www.hemaware.org/story/family-planning-carriers>
  - b. Guidance: This article provides some considerations for carriers as they decide to have children.
  - c. Limitation: This article may be somewhat outdated, as sperm sorting may no longer be an option for family planning in the United States.
12. **“What is Carrier Testing and Who Should be Tested,”** from The Hemophilia, von Willebrand Disease and Platelet Disorders Handbook on the Hemophilia of Georgia website
  - a. Download a copy at: <http://www.hog.org/handbook/article/2/12/what-is-carrier-testing-and-who-should-be-tested>
  - b. Guidance: This resource provides a short overview of carrier testing and who should be considered for testing.
13. **“What are Symptomatic Carriers?,”** from The Hemophilia, von Willebrand Disease and Platelet Disorders Handbook on the Hemophilia of Georgia website
  - a. Download a copy at: <http://www.hog.org/handbook/article/2/12/what-are-symptomatic-carriers>
  - b. Guidance: This resource provides a definition of a symptomatic carrier.

## II. Genetics of Hemophilia

1. **Inheritance of Hemophilia**, by the National Hemophilia Foundation
  - a. Order copies at:  
<http://www.hemophilia.org/NHFWeb/MainPgs/MainNHF.aspx?menuid=204&contentid=27>
  - b. Guidance: This resource is thorough and provides some valuable information.
  - c. Limitations: Some of the information is outdated. For instance, pre-implantation genetic testing is no longer considered “experimental”, sperm sorting may no longer be available in the U.S., and the likelihood of detecting a mutation in hemophilia A and B patients is not accurate based on current knowledge.
2. **Genetics Education Video**, from Virginia Commonwealth University HTC
  - a. 2nd link under Patient Information at:  
<http://www.vcuhealth.org/cvccd/resources/presentations.html>
  - b. Guidance: This video could be useful to some of your patients, particularly for visual learners.
  - c. Limitations: Some information is outdated. The video does not discuss symptomatic carriers. It also states that carrier status may influence insurance coverage or premiums, but does not address GINA.
3. **“How Hemophilia is Inherited,”** from The Hemophilia, von Willebrand Disease and Platelet Disorders Handbook on the Hemophilia of Georgia website
  - a. Download a copy at: <http://www.hog.org/handbook/section/2/how-hemophilia-is-inherited>
  - b. Guidance: This resource provides charts and explanations of the chances that children will be carriers or have hemophilia. This resource can also be used with other bleeding disorder patients.
  - c. Limitations: The description of a carrier may be a bit confusing.
4. **“How does a person get a bleeding disorder?,”** a page on the Steps for Living website
  - a. Download a copy at: <http://www.stepsforliving.hemophilia.org/basics-of-bleeding-disorders/genetics-of-bleeding-disorders/how-does-a-person-get-a-bleeding-disorder>
  - b. Guidance: This resource provides general inheritance information regarding hemophilia.
5. **“How Hemophilia is Inherited,”** a page on the CDC website
  - a. Download a copy at: <http://www.cdc.gov/ncbddd/hemophilia/inheritance-pattern.html>
  - b. Guidance: This resource also provides information on how hemophilia could be inherited in a family.
  - c. Limitations: The last example is somewhat misleading as it shows a 0% chance for child to have hemophilia, which is not totally accurate, as there is always chance for a new mutation.
6. **“What causes Hemophilia?,”** a page on the National Heart, Lung and Blood Institute webpage
  - a. Download a copy at: <http://www.nhlbi.nih.gov/health/health-topics/topics/hemophilia/causes.html>
  - b. Guidance: This resource provides information on how hemophilia is inherited.
7. **“Inheritance of Hemophilia,”** a page on the World Federation of Hemophilia website
  - a. Download a copy at: <http://www.wfh.org/en/abd/carriers/carriers-inheritance-of-hemophilia-en>
  - b. Guidance: This is another resource regarding the inheritance of hemophilia. There is also an explanation of lyonization.
8. **Indiana Hemophilia and Thrombosis Center, Bleeding Disorder Genetics Manual**
  - a. Available by request from Meadow Heiman at [mheiman@IHTC.org](mailto:mheiman@IHTC.org)

- b. Guidance: This resource is targeted for providers and it is a thorough overview of general genetics, the genetics of bleeding disorders, testing options and family planning options.

### III. Genetic Testing

1. **“Genetic Testing for Hemophilia,”** a page of the Cincinnati Children’s Hospital website
  - a. Download a copy at: <http://www.cincinnatichildrens.org/health/h/hemophilia-test/>
  - b. Guidance: This page gives brief explanations of various aspects of genetic testing, including linkage and direct mutation testing.
2. **“When to test for Carrier status,”** a page on the WFH website
  - a. Download a copy at: <http://www.wfh.org/en/abd/carriers/carriers-when-to-test-for-carrier-status-en>
  - b. Guidance: A brief look at the issues to consider when contemplating genetic testing for carrier status.
3. **“Test Takers,”** an article from NHF’s HemAware magazine
  - a. Download a copy at: <http://www.hemaware.org/story/test-takers>
  - b. Guidance: A comprehensive look at carrier testing from the ethical issues, to explaining how and when to test.
4. **“Hemophilia Genotyping for All,”** an article from NHF’s HemAware magazine
  - a. Download a copy at: <http://www.hemaware.org/story/hemophilia-genotyping-all>
  - b. Guidance: This resource advocates for the genotyping of patients for the improvement of clinical care.
5. **“The benefits of knowing your genotype,”** an article from NHF’s HemAware magazine
  - a. Download a copy at: <http://www.hemaware.org/story/your-genotype>
  - b. Guidance: This resource lists some of the advantages of genetic testing.
6. **“Genetic Counseling,”** an article from NHF’s HemAware magazine
  - a. Download a copy at: <http://www.hemaware.org/story/genetic-counseling>
  - b. Guidance: This article covers counseling issues, GINA and other questions that patients may have once they receive their genetic testing results.
7. **“Genetic Mutations 101,”** an article from NHF’s HemAware magazine
  - a. Download a copy at: <http://www.hemaware.org/story/genetic-mutations-101>
  - b. Guidance: This article reviews the types of mutations and what these can tell us about a patient’s risks.

### IV. Uses for genetic testing results

1. **“Genetic Differences Influence Inhibitor Development,”** an article found in NHF’s HemAware magazine
  - a. Download a copy at: <http://www.hemaware.org/story/genetic-differences-influence-inhibitor-development>
  - b. Guidance: This resource explains that certain types of genetic variations can be more prone to inhibitors. This may provide a motivation for patients to be tested, as more connections like these are made with further research.
2. **“Probing Gene Therapy,”** an article found in NHF’s HemAware magazine
  - a. Download a copy at: <http://www.hemaware.org/story/probing-gene-therapy>
  - b. Guidance: This resource provides updates in the field of gene therapy and research.