Genetic Conditions

PREFER CHW TRAINING

Empowering Communities through Genomics Education

Learning Objectives

- Understand and explain genetic conditions in community-friendly language
- Recognize common genetic conditions that may impact communities CHWs serve
- Describe the role of early detection and intervention in improving health outcomes
- Identify strategies for CHWs to educate and support patients and families regarding genetic conditions

PREFER CHW TRAINING

Empowering Communities through Genomics Education

What are Genetic Conditions?

Genetic conditions are health problems caused by changes (mutations) in a person's DNA

These changes can be inherited (passed down from parents) or happen by chance.

PREFER CHW TRAINING

Empowering Communities through Genomics Education

Types of Genetic Conditions

Single gene disorders:

- mutation in one gene causes the problem
- cystic fibrosis, sickle cell, Huntington's disease

Chromosomal disorders:

- a problem with the number or structure of chromosomes
- Down syndrome, Turner syndrome

Multifactorial disorders:

- caused by both DNA and things in the environment

PREFER CHW TRAINING
Empowering Communities through Genomics Education

Single Gene Disorders

Happens when there is a mutation in the DNA of one gene. These mutations can cause a range of health problems.

PREFER CHW TRAINING

Empowering Communities through Genomics Education

Single Gene Disorders

Tier 1 Conditions

- Type of single gene disorder
- Finding them early can help lower the chance of getting sick or make the illness less serious
- There are good ways to check for, prevent, and treat them
- About 1 in 75 people have a Tier 1 condition but many don't know they're at risk
- Finding people who are at risk can help stop serious health problems.

PREFER CHW TRAINING
Empowering Communities through Genomics Education

CHW Support for Single Gene Disorders

Teach	Teach people about these conditions
Help	Help connect people to the right care
Educate	Provide health education

PREFER CHW TRAINING

Empowering Communities through Genomics Education

Sickle Cell Disease

Single gene disorder that affects red blood cells.

Passed down from parents.

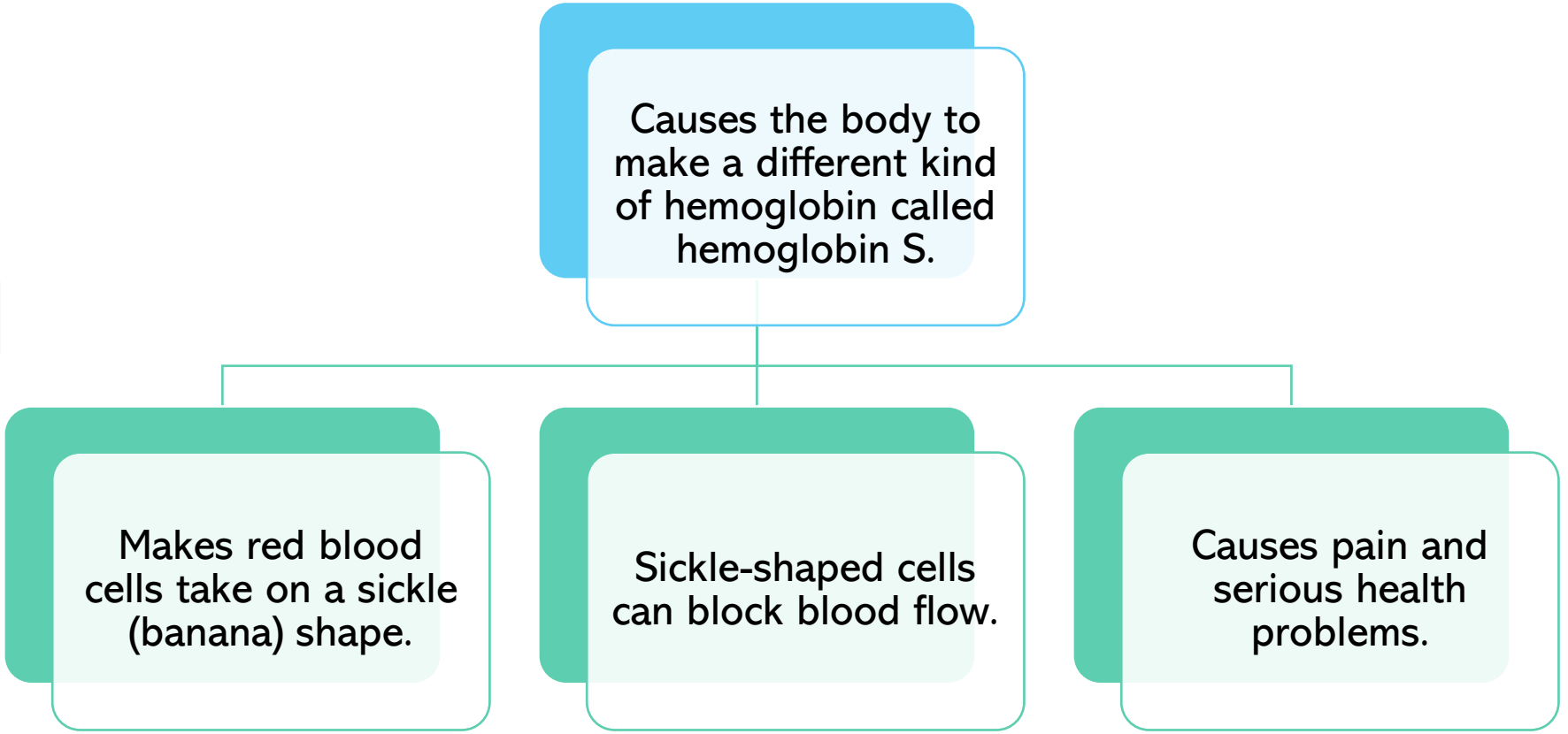
Caused by a mutation in the HBB gene

Must get **a copy** of the sickle cell gene from each parent to get the disease **(autosomal recessive)**

PREFER CHW TRAINING

Empowering Communities through Genomics Education

Sickle Cell Disease

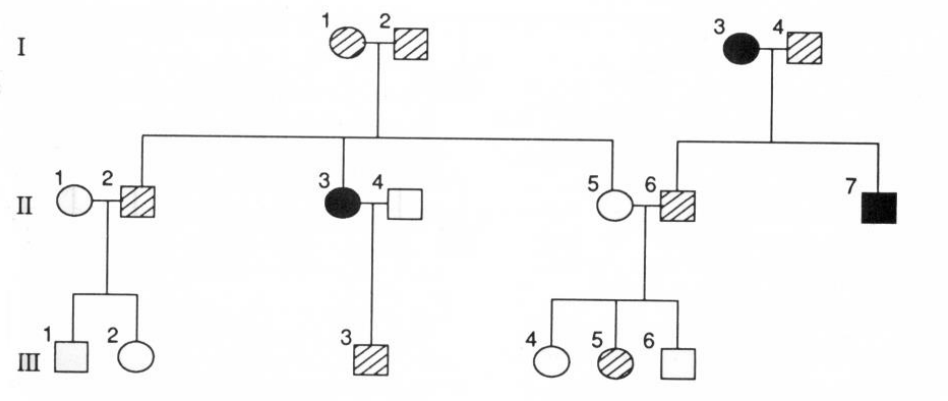


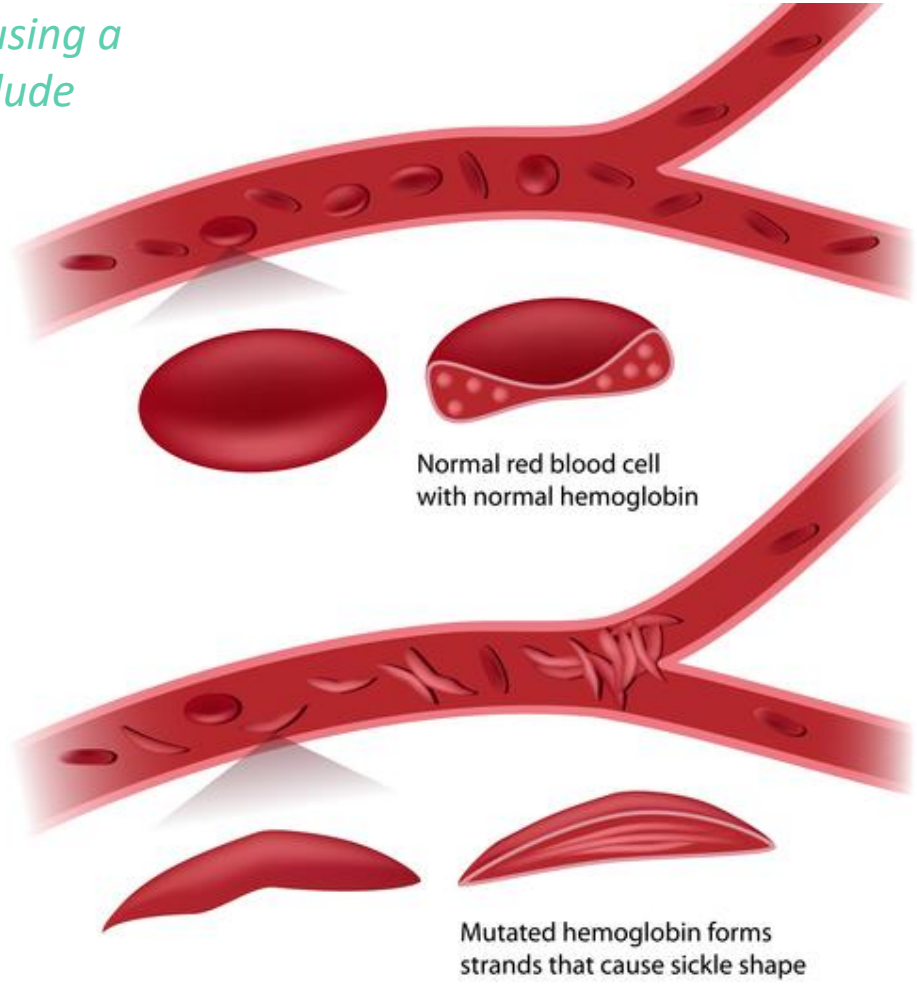
PREFER CHW TRAINING

Empowering Communities through Genomics Education

Sickle Cell Anemia (disease) and Sickle Cell Trait

To clearly show the inheritance of sickle cell disease and sickle cell trait using a pedigree, here is an explanation and a basic outline of what it might include





Hb^AHb^A = normal hemoglobin = normal red blood cells (RBCs)

Hb^AHb^S = sickle-cell trait = few sickled RBCs

Hb^SHb^S = sickle-cell anemia = many sickled RBCs

	Hb^A	Hb^S
Hb^A	Hb^AHb^A	Hb^AHb^S
Hb^S	Hb^AHb^S	Hb^SHb^S

PREFER CHW TRAINING

Empowering Communities through Genomics Education

CHW Support for Sickle Cell

Support	Support early testing and diagnosis
Teach	Teach families about the condition
Help	Help connect people to care and support

PREFER CHW TRAINING
Empowering Communities through Genomics Education

Hereditary Breast and Ovarian Cancer (HBOC) Syndrome

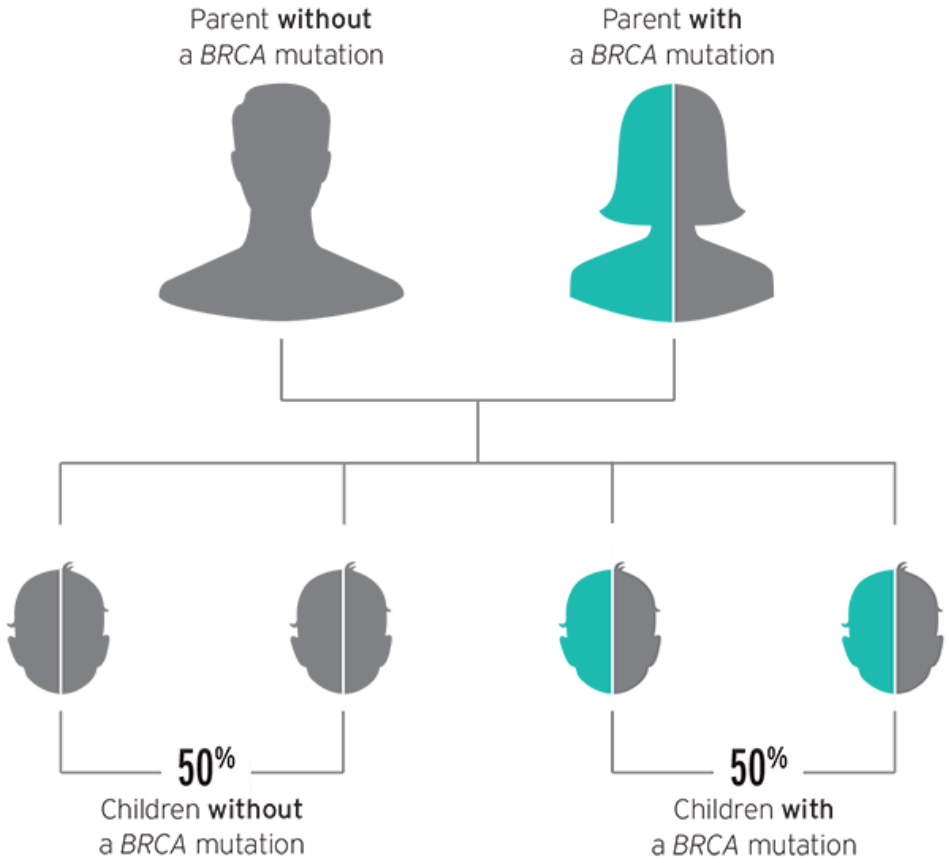
Pattern of cancer in families

- May result in higher-than-normal risk
- May be caused by mutations in genes that are passed in families

May present as:

- Same type or set of cancers in multiple family members
- Developing cancer at an early age
- Two or more types of cancer in the same person
- Example: breast and ovarian cancer

Risks for Hereditary Breast and Ovarian Cancer



Increased age

Genetic mutation (like BRCA1 and/or BRCA2)

Personal history of cancer

Reproductive history and infertility

Obesity

Family history of:

- Breast cancer (any type)
- Ovarian cancer
- Prostate cancer
- Pancreatic cancer

PREFER CHW TRAINING

Empowering Communities through Genomics Education

Risks for Hereditary Breast and Ovarian Cancer

BRCA1 can increase risk of:

- Cervical cancer
- Uterine cancer
- Colon cancer

BRCA2 can increase risk of:

- Stomach cancer
- Gallbladder cancer
- Bile duct cancer
- Melanoma

PREFER CHW TRAINING

Empowering Communities through Genomics Education

CHW Support for HBOC Syndrome

Educate	provide culturally relevant education
Facilitate	facilitate genetic counseling and testing
Guide	offer guidance on risk management

PREFER CHW TRAINING

Empowering Communities through Genomics Education

Lynch Syndrome

Increased risk of:

- Colorectal cancer
- Endometrial cancer
- Ovarian cancer
- Upper GI tract
- Urothelial cancer
- Prostate cancer
- Pancreatic cancer
- Brain cancer

Associated genes:
MSH6, MSH2, EPCAM, MLH1, & PMS2.

PREFER CHW TRAINING
Empowering Communities through Genomics Education

Lynch Syndrome Risk Factors

Personal history of colorectal cancer (especially those diagnosed before age 50)

Family History

- Multiple family members who have had colorectal, endometrial or ovarian cancer.
- Two generations of colon or rectal cancer
- At least one generation with colon or rectal cancer and one generation with polyps
- First-degree relative with colorectal or uterine cancer and another Lynch-syndrome related cancer at the same or at a different time.

CHW Support for Lynch Syndrome

Educate	Educate them on hereditary cancer risks
Promote	Promote regular screening and genetic counseling
Navigate	Help them navigate risk management options and family communication.

PREFER CHW TRAINING
Empowering Communities through Genomics Education

Familial Hypercholesterolemia (FH)

- An inherited condition of high levels of LDL cholesterol (“bad” cholesterol) in the blood.
- The liver doesn’t remove LDL cholesterol like it should
- Causes cholesterol to build up in the blood
- Genes linked to FH: LDLR (most common), APOB, & PCSK9

PREFER CHW TRAINING

Empowering Communities through Genomics Education

FH Health Risks

- Coronary artery disease (narrow or blocked heart arteries)
- Heart attack (also called myocardial infarction)
- Heart failure (when the heart can't pump well)
- Aortic stenosis (narrowing of a heart valve)

PREFER CHW TRAINING
Empowering Communities through Genomics Education

CHW Support for FH

Raise	Raise awareness about inherited high cholesterol
Promote	Promote early screening and treatment
Assist	Assist with lifestyle modifications
Support	Support Access to care

PREFER CHW TRAINING

Empowering Communities through Genomics Education

Chromosomal Disorders

- Occur when there is a change in the number or structure of chromosomes
- People usually have 23 pairs of chromosomes (46 total).
- A chromosomal disorder can happen when:
  - There is an extra chromosome
  - A missing chromosome
  - A change in the shape or structure of a chromosome
- Ex: Down Syndrome
  - Caused by an extra copy of chromosome 21
  - Can be found during prenatal screening (before a baby is born)

Chromosomal Disorders

Occurs when there is a change in the number or structure of chromosomes

People usually have 23 pairs of chromosomes (46 total).

A chromosomal disorder can happen when:

- There is an extra chromosome
- A missing chromosome
- A change in the shape or structure of a chromosome

Ex: Down Syndrome

- Caused by an extra copy of chromosome 21
- Can be found during prenatal screening (before a baby is born)

PREFER CHW TRAINING

Empowering Communities through Genomics Education

CHW Support for Chromosomal Disorders

Educate	Inform expectant families about prenatal screening and genetic testing

PREFER CHW TRAINING

Empowering Communities through Genomics Education

Multifactorial Disorders

Caused by a both genes and the environment

Often **run in families**, but pattern of inheritance is not simple

Things like **lifestyle, diet, weight, and age** can increase risk

Ex: Type 2 Diabetes caused by:

- Genetic factors (like family history)
- Environmental factors (like poor diet, obesity, and getting older

CHW Support for Multifactorial Disorders

Educate	Educate about the role of genetic and environmental factors
Encourage	Encourage screening for risk factors
Connect	Connect individuals to resources

PREFER CHW TRAINING
Empowering Communities through Genomics Education

Educating Patients and Families about Genetic Conditions

- **Explain genetic conditions** in a way that's easy to understand and respectful of different cultures
- **Help patients collect family health history** by showing them how to look for patterns and use simple tools like family history charts (more on this in the next module)
- **Encourage people to talk with their doctors** and explain the benefits of **genetic testing and counseling**

PREFER CHW TRAINING

Empowering Communities through Genomics Education

Educating Patients and Families about Genetic Conditions

- Give examples of questions they can ask during medical visits, like:
  - “Is this condition genetic?”
  - “Should I get genetic testing?”
  - “What does this test mean for my family?”

PREFER CHW TRAINING

Empowering Communities through Genomics Education

Educating Patients and Families about Genetic Conditions

Decide whether it's better to talk one-on-one or in a group based on:

- Privacy
- Cultural traditions
- Emotions around health and family

Clear up myths about genetic conditions, testing, and how traits are passed down

Share community resources, like:

- Free or low-cost testing
- Support groups
- Counseling services

PREFER CHW TRAINING

Empowering Communities through Genomics Education

Educating Patients and Families about Genetic Conditions

Help people overcome barriers like:

- Language
- Cost
- Accessibility

Talk about the power of prevention, including:

- Healthy lifestyle choices
- Medicines or treatments
- Steps to lower risk

PREFER CHW TRAINING

Empowering Communities through Genomics Education

Workbook Activity #2

Using the CDC Tier 1 Genomics Applications Tool Kit, complete the following scavenger hunt!
Go to: https://archive.cdc.gov/www_cdc_gov/genomics/implementation/toolkit/index.htm

1. Tier 1 genomic applications are those that have the potential for positive impact on public health based on evidence-based guidelines and recommendations.
   True
   False

2. **Fill in the Blank:**
   Nearly _____ people in the United States are at increased risk of Hereditary Breast and Ovarian Cancer, Lynch syndrome and/or Familial Hypercholesterolemia.

3. **Fill in the Blank:**
   - Hereditary Breast and Ovarian Cancer Syndrome (HBOC) increases the risk of breast, ovarian, tubal, peritoneal and other cancers due to mutations in the _____ or _____ gene.
   - Lynch Syndrome (LS) increases the risk of _____, endometrial, ovarian, and other cancers associated with mismatch repair genes.
   - Familial Hypercholesterolemia (FH) is associated with increased risk of _____ _____ or stroke due to mutations leading to very high _____ levels from an early age.

Workbook Activity #2

Reflection Questions

1. In your own words, write a simple definition of a genetic condition that you could use to explain it to a community member or client

2. Choose one common genetic condition (e.g., sickle cell, HBOC, Lynch syndrome, FH) and write how you would explain this condition in plain, culturally appropriate language. What visuals or tools could you use to help clients understand it?

3. What is one new thing you learned from this module that will help you in your CHW role?

4. What additional resources or support would help you feel more confident in educating patients and genetic conditions?