

Cascade Screening

What is cascade screening, and why is it important for identifying genetic conditions?

Cascade screening is a method used to identify individuals within a family who may be at risk of inheriting specific genetic conditions. It involves screening close relatives of a person diagnosed with a specific genetic condition to determine if they also have the same genetic mutation.

In the case of Familial Hypercholesterolemia (FH) and elevated Lipoprotein(a) {Lp(a)}, early intervention can help manage LDL (bad) cholesterol levels and reduce the risk of cardiovascular disease.

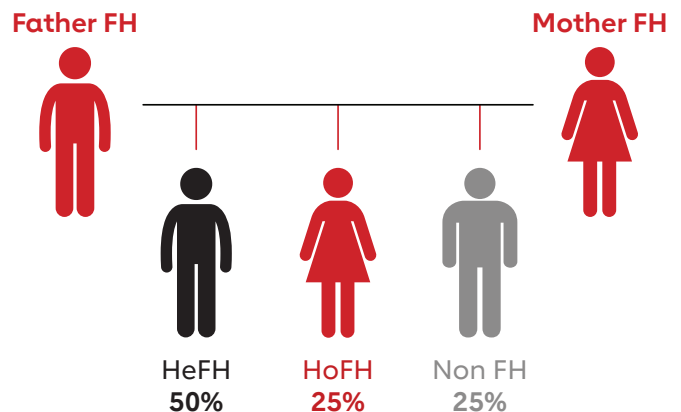
How does cascade screening work for identifying FH?

- 1) Identify person who has diagnosed FH.
- 2) Screen first-degree relatives including parents, siblings and children.
- 3) Encourage second-degree relatives and all family members to be screened.

What are the chances of family members having FH?

FH runs in families and can affect both children and adults. When both parents have FH, the chances of passing it on change, resulting in the following possibilities:

- 50% probability of inheriting HeFH, when a single gene is mutated.
- 25% probability of inheriting HoFH, when two genes are mutated.
- 25% probability of inheriting two unaffected genes that don't cause FH.



For Homozygous Familial Hypercholesterolemia (HoFH) is a rare and more severe form of FH inherited from both parents. If not detected and treated early, HoFH can lead to deadly cardiovascular complications in childhood.

Who should consider getting screened for genetic conditions like FH?

- First-degree relatives of a person with FH such as parents, siblings, and children of someone with FH.
- Those with a family history of FH.
- People with unexplained high cholesterol or at risk of having a heart attack or stroke.

Talk to your health care professional about cascade screening and other genetic testing options for your specific needs.

Learn more at heart.org/fh