Government of **Western Australia**
Department of **Health**

Royal Perth Hospital

Public forum on screening children for familial hypercholesterolaemia

All attendees are asked to have read this booklet ahead of the forum

Contents

Purpose of this booklet

The aim of this booklet is to provide you with some information to get you thinking about the public forum on screening children for familial hypercholesterolaemia (FH). It presents some of the issues that will be explored at the forum.

The booklet includes:

- information on FH
- information on what a screening program is
- some questions you might want to keep in mind when you come to the forum.

Some arguments for and against screening children for FH are included in this booklet. However, not all possible perspectives are included. We will provide other perspectives at the forum and you may have ideas of your own. We hope you will bring these to the forum.

Why you should be involved

Participating in this public forum offers a unique opportunity to help inform any decisions made by the WA Health on screening children for FH.

Please come to the forum:

- having read the information provided in this booklet
- ready to contribute
- open to the ideas and opinions of others.

About the public forum

Doctors and policy makers from WA Health have organised a public forum to discuss whether it is acceptable to screen children for FH. It's important that we find out what citizens think. People from all across WA have been invited to participate in this public forum. The goal is to find out whether members of the public believe a program that screens children for FH is acceptable. This goal will be achieved by bringing you together with people from different backgrounds, experiences and opinions, to learn about and discuss screening children for FH.

What you will be asked to do

At the forum, you will be asked to do three things.

- Listen to some presenters who will talk about different views and perspectives on screening children for FH
- Discuss with the other participants the information presented and each other's views on the issues
- Reach a group decision on whether you think screening children for FH is acceptable and if so, at what age children should be screened.

It doesn't matter if you don't know anything about FH and screening. You will be provided with all the information you need to be able to participate in the forum.

Why have a public forum?

Screening children for FH is a complex topic. While there are benefits to screening people for a condition, it can also result in harms and costs. This means screening programs must be considered very carefully before being established.

Firstly, there must be good scientific evidence that shows that it is safe and appropriate to screen for a condition. Secondly, a range of perspectives must be considered. Scientists, doctors, lawyers, ethicists, policy makers, interest groups and industry may all have different views on whether screening is acceptable. Some perspectives may include:

- the general public (citizens)
- the people who would be invited to participate in a screening program
- doctors and nurses, who would carry out tests and make sure that people receive appropriate care
- governments, who fund and operate the programs.

It is important to consider all perspectives before deciding whether to screen children for FH. The forum aims to understand the views and perspectives of the general public (citizens) of WA.

Familial hypercholesterolaemia

Familial hypercholesterolaemia (FH) is a condition that can result in high cholesterol levels in the blood. This can have very serious impacts on the health of a person with FH.

What is cholesterol?

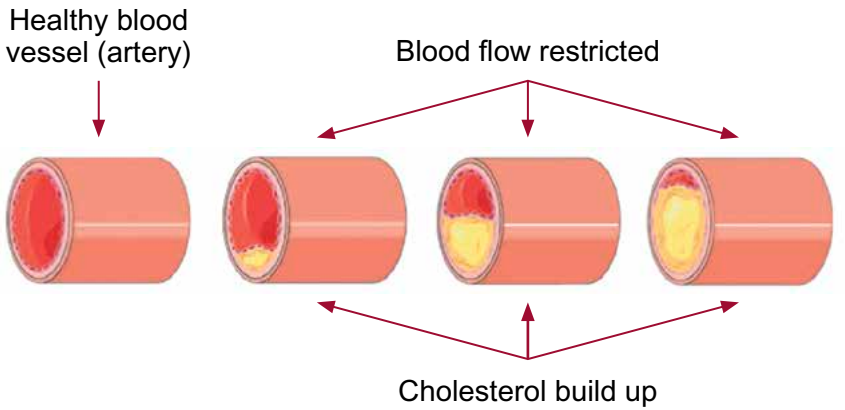
Cholesterol is a fatty substance found in the blood. The body needs cholesterol to make hormones and vitamin D, and to aid digestion. The body also uses cholesterol to build and maintain the outer wall of cells. Cholesterol is made by the body, and is also found in many foods, particularly eggs and other animal products. Saturated and trans fats also have cholesterol. The cholesterol from our diet is absorbed from the food we eat into the bloodstream.

While cholesterol is essential for the body to function properly, it is important that the amount of cholesterol in the blood does not get too high.

Can cholesterol be harmful?

If there is too much cholesterol in the bloodstream, it can stick to the walls of the blood vessels. As shown in Figure 1, this causes the blood vessels to narrow and they may eventually become blocked.

Figure 1: High blood cholesterol leads to cholesterol build up in a blood vessel wall.



Courtesy of Servier Medical Art

This will affect blood flow to the heart and other vital organs, and can cause heart attacks, strokes and other problems. A poor diet, diabetes, lack of exercise and obesity all increase the risk of high cholesterol and heart disease. Heart disease has been the leading cause of death in Australia since 2000, causing 13.4 per cent of all deaths in 2013.

What is FH?

Familial hypercholesterolaemia literally means “high cholesterol in a family”. People with FH have high cholesterol levels and an increased risk of heart disease.

In Australia, about one in every 300 people has FH.

How do you get FH?

FH is inherited from one or both of your parents. Each parent gives you one copy of the gene that helps control the amount of cholesterol in the blood. Sometimes the instructions given by the gene are faulty. If a parent has FH, there is a one in two chance that their child will also inherit FH.

What happens to people with FH who are not diagnosed?

People with FH who are not diagnosed cannot be treated. This means they have a 100-fold increase in the risk of heart disease, with 50 per cent of males and 20 per cent of females with FH suffering a heart attack or stroke by the age of 60.

What can be done about FH?

Once diagnosed, FH is relatively easy to treat. People with FH may be able to avoid getting heart disease by leading a healthy lifestyle, along with taking medications prescribed by their doctor. A healthy lifestyle includes eating a healthy diet, not smoking, exercising regularly, staying at a healthy weight and enjoying alcohol in moderation.

While lifestyle is important, it rarely does enough in people with FH to lower cholesterol to the recommended levels. This means medication, usually with drugs called statins, is required for almost all people with FH.

When should treatment for FH start?

The use of cholesterol-lowering treatments, such as statins, should be guided by the person's age, the family history of cardiovascular diseases such as heart attacks and strokes, the level of cholesterol, as well as the views of the individual and their family. Current guidance from clinicians around the world suggests that treatment with statins should begin between the ages of eight and 10 years of age in both boys and girls with FH.

Diagnosing FH in WA

Adults may be diagnosed with FH after going to hospital with a heart attack or stroke, or after visiting their GP and finding out they have a high blood cholesterol level.

There is a government program in WA, called FHWA, which aims to find new cases of FH in families. It does this by testing the relatives (including children) of patients newly diagnosed with FH, to see if the relatives also have FH. This is called cascade screening.

Cascade screening is a cost-effective way to find new cases of FH. However, FHWA has been running since 2009 and overall has found around 800 people with FH, which is about 10 per cent of the estimated 8,000 people with FH in WA.

It is thought that around 1,500 people with FH in WA are under 16 years of age. However, only 71 of these 1,500 children have been identified through the FHWA program. This means more than 95 per cent of children with FH in WA remain undiagnosed.

The diagnosis rates in WA are similar to those in other developed countries, such as the United Kingdom.

The low number of people that have been diagnosed with FH, compared to the expected number of people in WA with FH, means that new ways to identify those people living with FH need to be explored.

One of the options being thought about by WA Health for diagnosing children with FH is called universal population screening.

What is universal population screening?

Universal population screening is where a test for a condition is offered, in an organised way, to all people in a defined population. This helps to identify people with a higher chance of having the condition. People who receive a positive screening result can then be referred to specialists to receive a diagnosis.

The goal of universal screening is to find people with a condition before they display any symptoms, so that they can receive treatment that limits the condition's effect on their health. This then reduces the amount of illness and death in the population caused by the condition. A number of successful screening programs currently run in WA, including BreastScreen WA and Newborn Bloodspot Screening (known as the heel prick test for newborn babies).

Under a universal screening program for FH, every child would receive an offer to have their cholesterol levels tested when they reached a certain age. This involves taking a finger-prick blood test. The results would tell people whether they are at risk of FH and if so, they can then go to a specialist to get a diagnosis. This would hopefully increase the number of children diagnosed with FH.

When could screening occur?

Guidance from doctors around the world suggests that treatment for FH, using statins, should begin from the age of eight to 10 years. This may be a suitable time to offer universal population screening of children for FH.

There are also a number of other possibilities for how a universal screening program could be offered. One option is to offer a blood test to measure cholesterol levels at the same time as when a child receives their immunisations. This may be at either:

- four years of age, at the time of the pre-school booster, or
- 12–13 years of age, at the time of the year 8 HPV vaccine.

Combining FH screening with childhood immunisations lowers both program costs and patient inconvenience, because the child does not need to make a separate appointment to take the test. However, it may also have a negative impact on participation in the immunisation programs. If parents do not wish for their child to be screened, they may refuse both FH screening and immunisations.

Is screening the best choice?

Screening all children for FH may not be the most appropriate way to try and increase the diagnosis of FH. Other options include:

- continuing with the current cascade screening approach. So far, this has not had much of an impact on increasing the number of people diagnosed with FH.
- increasing the awareness of FH, among both the public and General Practitioners (GPs). An education program could be established so that more people know about FH and hopefully more people will go to their GP for testing. However, this will likely result in a lower rate of diagnosis than universal screening.

There may be other options that are presented at the forum and you may have your own ideas about this.

Things to consider for screening

A screening program needs to be considered very carefully before it is established, to make sure that it will be safe, effective, equitable (that is, fair and available to all children in WA) and worthwhile.

Screening programs are very complex and expensive to run, so it is vital that they are based on evidence. Some things

WA Health would need to consider in deciding whether to screen children for FH would include:

- Is FH an important and serious health problem?
- Do the benefits of screening outweigh the harms and costs?
- Is the blood test safe and able to accurately detect every child with FH?
- Is there an acceptable treatment for children diagnosed with FH?
- How can the test be made available to every child in WA?
- Are there enough staff and facilities to offer testing?
- Is screening the best option to increase diagnosis?

The answers to these questions will be explored at the forum and you might like to keep them in mind on the day.

In addition, because government resources can be used to fund and run a screening program, it should be clear whether the public finds screening children for FH acceptable. That is why the public forum is being held.

Arguments for screening children

Benefits of screening children for FH include earlier diagnosis and treatment of FH. This may reduce their risk of getting heart disease in adult life.

Damage to arteries in people with FH begins at a young age. It is important that people with FH are diagnosed early, to make sure that they are able to receive treatment before permanent damage to their arteries is done. However, current diagnosis rates are very low. Universal screening offers a way to greatly increase the number of people diagnosed with FH, and decrease the huge impact of heart disease in WA.

Blood tests to measure cholesterol levels are relatively cheap. After receiving a diagnosis, treatment of FH with cholesterol-lowering medication such as statins is safe and effective.

When a child is diagnosed with FH, it also means that their parents can be tested for FH. This means parents could start treatment for FH before they suffer a heart attack or stroke.

Arguments against screening children

If a child is diagnosed with FH, then they must have inherited it from their mother or father. If both parents are tested for FH and neither have the condition, it may raise questions about the child's biological parents.

There is some disagreement between researchers on the accuracy of blood tests for identifying people at risk of FH. It is important that tests are accurate. If the test incorrectly tells someone they are not at risk of FH, this may result in a heart attack or stroke later in life. On the other hand, children that do not have FH, or have a mild form, may be diagnosed and treated when they may never have suffered symptoms or required medication. The test has been found to be most accurate in children between the ages of one and nine.

There is always a level of pain and anxiety associated with a blood test, particularly in younger children.

A diagnosis of FH could potentially affect a person's insurance premiums.

Very little research has been done on how much a universal screening program for FH would cost. This makes it difficult to determine whether the benefits of screening for FH outweigh the costs. It is also hard to say if universal screening is the most cost-effective option to increase diagnosis rates for FH.

The most accepted way to see whether a screening program would be effective is to first trial it on a random sample of the population. To date, no large scale randomised trials of screening for FH have been reported.

Your decisions to make

At the public forum, you will be asked to listen to a range of opinions and perspectives on screening children for FH, and as a group decide two things:

1. Is screening children for FH acceptable?
2. If so, at what age should screening be offered to children?

This booklet is designed to provide you some background information to help you make these decisions.

For more information

If you have any questions or require further information about the contents of this booklet or the forum itself, please contact the Forum Coordinator:

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Notes

Notes

Arguments for and against screening children for FH

The condition (FH)

The test

The treatment

The screening program

This document can be made available in alternative formats on request for a person with a disability.

Produced by the Office of Population Health Genomics

GEN-013078 JAN'16