



Video transcript

Introduction to Mackenzie's Mission

Mackenzie's Mission is an Australian Government funded research study.

It aims to understand how best to offer a test called reproductive genetic carrier screening. Genetic screening gives parents planning a pregnancy, or early in pregnancy, information about their chance of having a child with a serious genetic condition. Thousands of couples across Australia will be invited to take part in this study by their healthcare provider.

The study is named after our daughter Mackenzie, who passed away at seven months of age from an inherited genetic condition called spinal muscular atrophy. Like us, most parents who have a child with a genetic condition have no idea they have a risk until their child is born.

Our genes provide instructions for our bodies to develop and function. We all carry changes in some of these genes; usually, these changes don't cause any problems. However, sometimes the changes can make the gene faulty and can cause genetic conditions. Reproductive genetic carrier screening can detect some of these genetic faults, providing people with information about the chance of genetic conditions in their children.

Screening through the study will tell us whether a couple together have an increased chance of having a child with a serious childhood onset condition. Screening is free, simple and optional.

We are interested to hear from people who know that they want to have screening, are unsure, or know that they don't. Taking part involves completing at least one online survey and for those who choose to have screening, providing a mouth swab sample. Both members of the couple return their mouth swab samples via mail.

Results will tell the couple whether they have a low or increased chance of having a child with any of the genetic conditions screened. People who find out that they have an increased chance will be offered information about reproductive options and support in making choices based on their own values.

Screening is available to couples planning a pregnancy and also to those early in pregnancy. Couples will be invited by a participating healthcare professional. In order to participate, both biological parents need to be available, need to be over the age of 18 and the woman needs to be less than 10 weeks pregnant.

If a sperm, egg or embryo donor has or will be used for the pregnancy, having screening as part of this study is possible if the donor is willing to take part.

Researchers will assess the outcomes of screening, the experience of participants and healthcare providers, as well as ethical and health economic factors. This will provide important information on how to offer reproductive genetic carrier screening as a national program.

More information is available on the study website: mackenziesmission.org.au