Facts for people and families with Peutz-Jeghers syndrome

ID: 3431 v.3 **Endorsed**

This fact sheet contains general information about Peutz-Jeghers syndrome (PJS). Each person should be referred to a genetic service for further information and advice.

Key Points

- PJS is caused by faults (mutations) in the STK11 gene.
- Both males and females with PJS have an increased chance of developing cancer of the gastrointestinal tract, stomach and pancreas. Most people develop skin pigmentation and polyps (hamartomas) of the gastrointestinal tract.
- Females with PJS have an increased chance of developing breast, cervical and ovarian cancer.
- Family members (including children) can be referred to a genetic service where experts can provide information, advice and support about their chance of developing cancer and the option of genetic testing.

What is a faulty STK11 gene?

STK11 is a 'cancer protection' gene that helps to protect against cancer of the gastrointestinal tract, pancreas, breast, cervix and ovary.

Everyone has two STK11 genes (one from their mother, and one from their father). If one of the genes is not working, this is known as having a *faulty* STK11 gene, or having an STK11 *mutation*. People with a faulty STK11 gene are known to have Peutz-Jeghers syndrome.

What is the risk of cancer and other features of PJS?

Cancer:

- Both males and females with PJS have about a 40% chance of developing cancer of the large bowel and about a 15% chance of developing cancer of the small bowel over their lifetime.
- Both males and females with PJS have about a 30% chance of developing stomach (gastric) cancer over their lifetime.
- Both males and females with PJS have about a 25% chance of developing pancreatic cancer over their lifetime.
- Females with PJS have about a 45% chance of developing breast cancer and about a 20% chance of developing cervical cancer (adenoma malignum) or ovarian cancer (sex-cord stromal tumour) over their lifetime.
- Males with PJS have an increased chance of developing testicular tumours in childhood and adolescence.
- **Not everyone with PJS will develop cancer.**

Other features:

- Most people with PJS develop polyps in the gastrointestinal tract. Polyps can develop in childhood and may cause intussusception (where part of the bowel folds in on itself), bowel blockage, bleeding and/or abdominal pain.
- Most children with PJS develop specific skin pigmentation with dark blue/brown spots on the lips, inside the mouth and near the eyes and nostrils. This skin pigmentation does not cause health problems and usually fades with time.

How can this increased risk of cancer and gastrointestinal polyps be managed?

Gastrointestinal polyps:

- To **find gastrointestinal polyps and prevent complications**, people with PJS should have their stomach, small bowel and large bowel checked for polyps every 3 years, from age 8 years. This involves having a video capsule or magnetic resonance endoscopy, gastroduodenoscopy and colonoscopy. This screening may occur more or less frequently depending on whether polyps are identified or not. People with PJS should also have a blood test to check their haemoglobin every year.
- Children and adults with PJS should see a doctor if they develop severe abdominal pain, vomiting and/or bleeding from the bowel. These may be symptoms of intussusception requiring urgent medical attention.

Cancer in males and females:

- To **find bowel cancer early**, people with PJS should have bowel cancer screening at least every 3 years from age 30 years. This screening involves having a gastroduodenoscopy and colonoscopy. The aim is to find bowel cancer early in addition to preventing polyp related complications.
- **There is no reliable method of screening for pancreatic cancer.**
- To **reduce the chance of getting pancreatic cancer,** people with PJS should not smoke.

Cancer in females:

- To **find breast cancer early**, females with PJS should have breast cancer screening every year from age 30 years. This involves having a breast MRI (and sometimes a mammogram), plus a breast check by a doctor.
- To **reduce the chance of getting breast cancer,** females with PJS may take medications such as tamoxifen or raloxifene. Some females may consider breast surgery (risk-reducing mastectomy or RRM).
- **There is no reliable method of screening for ovarian cancer.**
- To **find cervical cancer early**, females with PJS should have their cervix screened every year from age 18 years. This involves having a pelvic examination and an endocervical smear with a specialist gynaecologist.

Cancer in males:

- To **find testicular tumours early**, males with PJS should have a physical examination (including testicular examination) with their doctor every year from birth through to their teenage years.

What does this mean for family members?

Family members (including children) of someone with a faulty STK11 gene can have genetic testing to check who has the faulty gene and who does not. Their doctor can refer them to a genetic service to find out more about their chance of developing cancer and what genetic testing involves.

If a person **does have** the faulty STK11 gene:

- they can pass it on to their children
- each child has a 50% (1 in 2) chance of being born with it. Pregnancy planning options are available to people who want to prevent the faulty gene being passed on.

If a person **does not have** the faulty STK11 gene:

- they have the same chance of developing cancer and other features of PJS as the general population (unless there are other factors that increase this risk)
- they cannot pass it on to their children.

People who decide not to have genetic testing should still get advice about managing their chance of developing cancer.

More information and support

- Centre for Genetics Education NSW Health: Contact details for local genetics services –genetics.edu.au/SitePages/Genetic-Services.aspx
- Genetic Alliance Australia – geneticalliance.org.au
- Cancer Australia – canceraustralia.gov.au

- Australian Pancreatic Cancer Genome Initiative – pancreaticcancer.net.au
- Breast Cancer Network Australia (BCNA) – bcna.org.au
- COSA - Medications to lower the chance of breast cancer: information for women – cosa.org.au/groups/cancer-genetics/resources/

History

Version 3

Date	Summary of changes
09/12/2019	Document reviewed in line with protocol ID 395 following May 2019 CG reference committee meeting. Approved for publication with the following changes made: • **Key points:** stomach cancer added • **What is the risk of cancer and other features of PJS?** section ◦ Bowel cancer and pancreatic cancer separated out into different bullet points ◦ Added "Both men and women with PJS have about a 30% chance of developing stomach (gastric) cancer over their lifetime." ◦ Added "Males with PJS have an increased chance of developing testicular tumours in childhood and adolescence." • **How can this increased risk of cancer and gastrointestinal polyps be managed?** section ◦ Added "To find testicular tumours early, men with PJS should have a physical examination (including testicular examination) with their doctor every year from birth through to their teenage years." • "Men" changed to "males" and "women" changed to "females" throughout document, as consumer sheet is applicable for adults and children Updated to version V.3. To be reviewed again alongside protocol ID 395 when next due for review.
12/05/2021	More information and support: Link to COSA - Medications to lower the chance of breast cancer: information for women inserted

Version 2

Date	Summary of changes
27/04/2018	Developed in conjunction with consumer information sheet working group and presented at the November 2017 Cancer Genetics meeting. Discussions continued over email and document approved for publication. V.1.
30/05/2019	Document reviewed in line with protocol ID 395 following November 2018 CG reference committee meeting. Approved for publication with the following changes made: **How can this increased risk of cancer and gastrointestinal polyps be managed?** section: • Surveillance frequency for stomach, small bowel and large bowel added. • Breast cancer surveillance recommendations updated. Version updated to V.2. To be reviewed again alongside protocol ID 395 when next due for review.

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