

## **Your comments and concerns**

For advice, support or to raise a concern, contact our Patient Advice and Liaison Service (PALS). To make a complaint, contact the complaints department.

**t:** 020 7188 8801 (PALS) **e:** [pals@gstt.nhs.uk](mailto:pals@gstt.nhs.uk)

**t:** 020 7188 3514 (complaints)

**e:** [complaints2@gstt.nhs.uk](mailto:complaints2@gstt.nhs.uk)

## **Language and accessible support services**

If you need an interpreter or information about your care in a different language or format, please get in touch.

**t:** 020 7188 8815

**e:** [languagesupport@gstt.nhs.uk](mailto:languagesupport@gstt.nhs.uk)

## **NHS website**

Online information and guidance on all aspects of health and healthcare, to help you take control of your health and wellbeing.

**w:** [www.nhs.uk](http://www.nhs.uk)

## **Become a member of the Trust**

Members of Guy's and St Thomas' NHS Foundation Trust contribute to the organisation on a voluntary basis. We count on them for feedback, local knowledge and support. Membership is free and it is up to you how much you get involved. To find out more, please get in touch.

**t:** 0800 731 0319 **e:** [members@gstt.nhs.uk](mailto:members@gstt.nhs.uk)

**w:** [www.guysandstthomas.nhs.uk/membership](http://www.guysandstthomas.nhs.uk/membership)

**Leaflet number: 4756/VER2**

Date published: June 2019

Review date: June 2020

© 2019 Guy's and St Thomas' NHS Foundation Trust  
A list of sources is available on request

## **How can genes and chromosomes cause disease?**

If a gene is mutated (changed) then the 'instruction' that it carries may not be read properly. This can cause disease. Usually, if someone has a gene mutation then this is found in every cell of the body.

If some chromosome material is missing, or extra, this can cause an imbalance of genetic messages. This can affect development. If someone has a chromosome mutation, this is usually found in every cell of the body.

## **How genetic and chromosomal diseases are usually passed on**

Many genetic and chromosomal diseases are passed down through families. For this to happen, one parent must be affected (dominant conditions), or both parents must be carriers (in recessive conditions). In other conditions the mutated gene is on the X chromosome so these diseases are described as being X-linked. Separate leaflets give more information about dominant, recessive and X-linked inheritance.

Sometimes a genetic or chromosomal disease happens 'de novo'. This means it occurs for the first time in a family when a baby is conceived and is not due to mutated genes or chromosomes in either parent. The chance that this random event will happen again to the same parents is usually very low. There may be a small possibility of having another affected pregnancy if one parent has germline mosaicism.