

# Creating patient information leaflets on mitochondrial diseases for adult patients

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## Introduction

**Mitochondrial disease is an umbrella term for a range of diseases caused by a dysfunctional mitochondria. Due to the complexity of the disease and the lack of awareness, it has been difficult to create accessible information for patients with mitochondrial disease.** This is something to be addressed about because patients need to understand the information in order to make informed decisions. Some patients also experience anxiety due to lack of information, which would impede their recovery. Although there are a few charities addressing this problem in children, most notably the Lily Foundation, there is significantly less information targeted to adults. The majority of mitochondrial patients at the Newcastle Mitochondrial Clinic are adults and are the target audience for the leaflets we created.

This is an ongoing project and the aim at the end is to produce a well-written and effective information leaflet that can be distributed to the patients at the Mitochondrial Clinic.

## Aims

1. To better understand how to effectively communicate information on mitochondrial diseases to patients and the general public
2. To create an accurate and accessible patient information leaflet that can be used as a template for future patient information leaflets

## Method

1. **Evaluation of current and available information.** An analysis was carried out comparing the information contents of several mitochondrial disease websites, including the Wellcome Trust's own. Several aspects were examined, including the relevance of the information and how the information was delivered.
2. **Creating the leaflet.** Using conclusions from the analysis, the author set out to create a draft leaflet on single large-scale deletions, using literature.
3. **Review by health professionals.** Before piloting it out to the patients and the public, the leaflet was sent to some health professionals within the mitochondria clinic.
4. **Piloting the leaflet to patients and public.** The draft leaflet was sent out to a patient focus group and VoiceNORTH, along with a questionnaire to evaluate it.

## Results

1. Evaluation of current and available information: Too much jargon leading to readability issues, font size too small, misleading information, lack of diagrams, relevance of information and difficulty navigating around the information.
2. The leaflet created is shown in figures 1 and 2
3. Pilot leaflet was sent to 9 health professionals, including a GP from Australia. Some of the comments included simplifying some sections and adding diagrams.
4. The leaflet has been sent to patients for feedback and replies are pending.

## Conclusions

The most important aspect of providing information for patients is to help reassure them about the prognosis of their disease. It was important that the information was written in a manner that emphasised hopes of recovering, without sacrificing accuracy. Observations from some of the patients show that anxiety regarding the disease can be debilitating, even more so because of how little known mitochondrial diseases are.

The second aspect highlighted is assuming patients understand commonly-used scientific terms such as 'gene' and 'DNA'. As these are difficult to explain in words, we thought diagrams would be the best way to clearly explain them (figure 3).

Sending the leaflets to patients would give us a better indicator of the effectiveness of our leaflets and would be the next best step with this project.

**Diagnosis**  
A muscle biopsy is often necessary. In adults, a small muscle sample can be taken from the shin or front thigh muscle using local anaesthetic. Scientists would look for changes in the muscle suggestive of abnormal mitochondria and undertake further testing to identify if there is deletion in the mitochondrial DNA. Urine and blood samples may also be taken.

**Management**  
There are effective treatments available to manage the symptoms and complications. For examples, insulin can be given to treat diabetes, a pacemaker inserted to control the heart rhythm and eyelid surgery to improve eyesight. Maintaining a healthy lifestyle is important so eating healthily and exercising regularly will be beneficial. You are encouraged to discuss with your doctor about the treatment and follow up that are relevant to you in the clinic.

**Contact**  
NHS Highly Specialised Service for Rare Mitochondrial Disorders  
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Tel 0191 2820840  
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Visit our website for further information  
www.newcastle-mitochondria.com

**Outcome**  
The long-term outcome of single, large scale deletions largely depends on what age the disease begins. Most patients with deletion disease will have a normal life-span and good quality of life. As we are getting better with medical treatments many patients even with severe disease live much longer. Researchers in Newcastle have recently developed a tool to predict how these diseases in individuals will likely progress, giving patients a better picture of the long-term outcome.

**Figure 1** Front cover of the single large-scale deletion leaflet

Text is large and easy to read

Key points highlighted in green

Diagram helps to visualise concepts

Large titles make it easier to navigate around the leaflet

Front cover is colourful and eye-catching

Contact section is clear and positioned for easy reference

**Mitochondrial diseases happen because of mutations in DNA.** A mutation is like a spelling mistake in the genetic code. In single large-scale deletion, a chunk of the mitochondrial DNA (mtDNA) is removed. When this happens, mitochondria cannot generate energy from food properly. As a consequence of energy deficiency, cells in various organs malfunction, leading to a variety of symptoms.

**Symptoms**  
The most common manifestation of single deletions is weakened eye muscles (ophthalmoplegia) and droopy eyelids (ptosis) in adults. Mitochondria are found in all cells and therefore any organ in the body can potentially be affected especially those with high energy-demand such as muscle, heart and brain.

**Syndromes**  
Chronic Progressive External Ophthalmoplegia (CPEO) usually manifests in droopy eyelids and difficulty moving the eyes. It may just involve weakening eye muscles but may also include other muscles such as the limbs and face. CPEO is a common symptom in patients with single deletions. Kearns-Sayre Syndrome (KSS) usually starts in patients under 20 years old. CPEO is a common sign, as well as a buildup of pigments in the retina (the back of the eye) that may affect night vision. Other signs and symptoms include general muscle weakness, difficulty balancing and slow heart rate (heart block). Pearson's Syndrome is a rare and severe disease occurring in infants. The main signs are a type of anaemia and pancreatic failure resulting in problems such as diabetes and malabsorption of food.

**Inheritance**  
The majority of single large-scale deletions are sporadic. This means that they are not passed on, but happen by chance. Variations in manifestations mainly depend on heteroplasmy (the proportion of mtDNA with mutation) and which chunk of DNA is removed.

Other signs and symptoms which may occur include:  

- Fatigue and exercise intolerance
- General muscle weakness
- Balance and coordination problems (ataxia)
- Heart involvement (slow heart rate that may result in blackouts)
- Diabetes mellitus and other hormone disorders

 Signs and symptoms that occur together are often grouped into syndromes.

**Figure 2** Back cover of the single large-scale deletion leaflet

Syndromes only list the common symptoms to give patients a brief overview

Names of the syndromes are sized according to their occurrence

## References

1. Glaser R. and Kiecolt-Glaser JK. 2005. Stress-induced immune dysfunction: implications for health. *Nature Reviews Immunology*. 5. pp243-251

Figure 3 Diagram visualising concepts such as genes and DNA