# PD Frontline Registration Guidance

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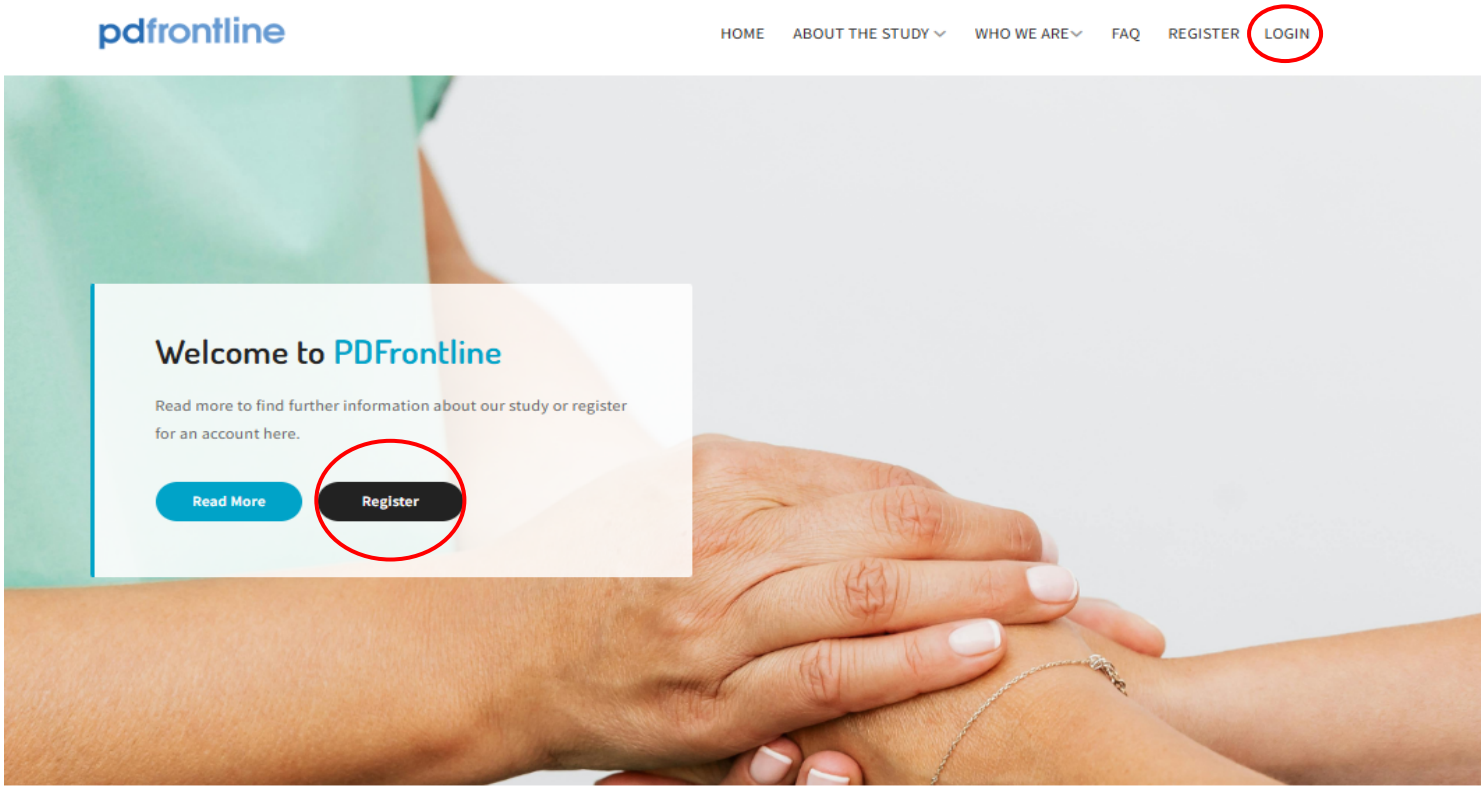
**Signing up**

To get started, you need to first sign up to PD Frontline. Please see guidance for this below:

1. Go to https://pdfrontline.com/en and select **register** complete the set of qualifying questions, or login to continue registration

   To successfully register to PD Frontline, you will need to select:

   1. **'YES'** to Do you live in the UK,
   2. **'NO (I don't know)'** to Do you carry a GBA mutation?
   3. **'YES'** to Do you have a confirmed diagnosis of Parkinson's Disease from a Clinician?

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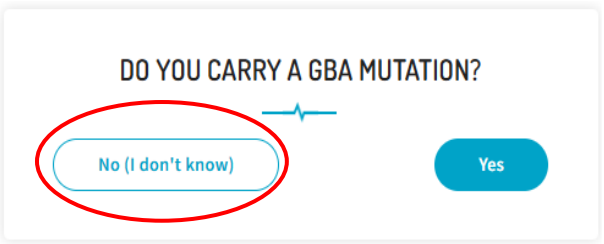


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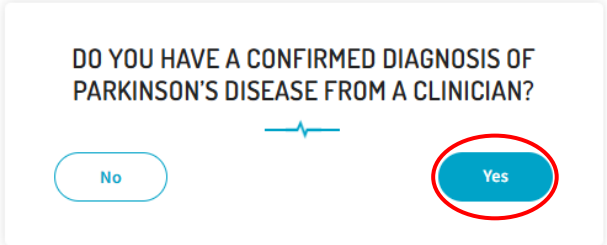
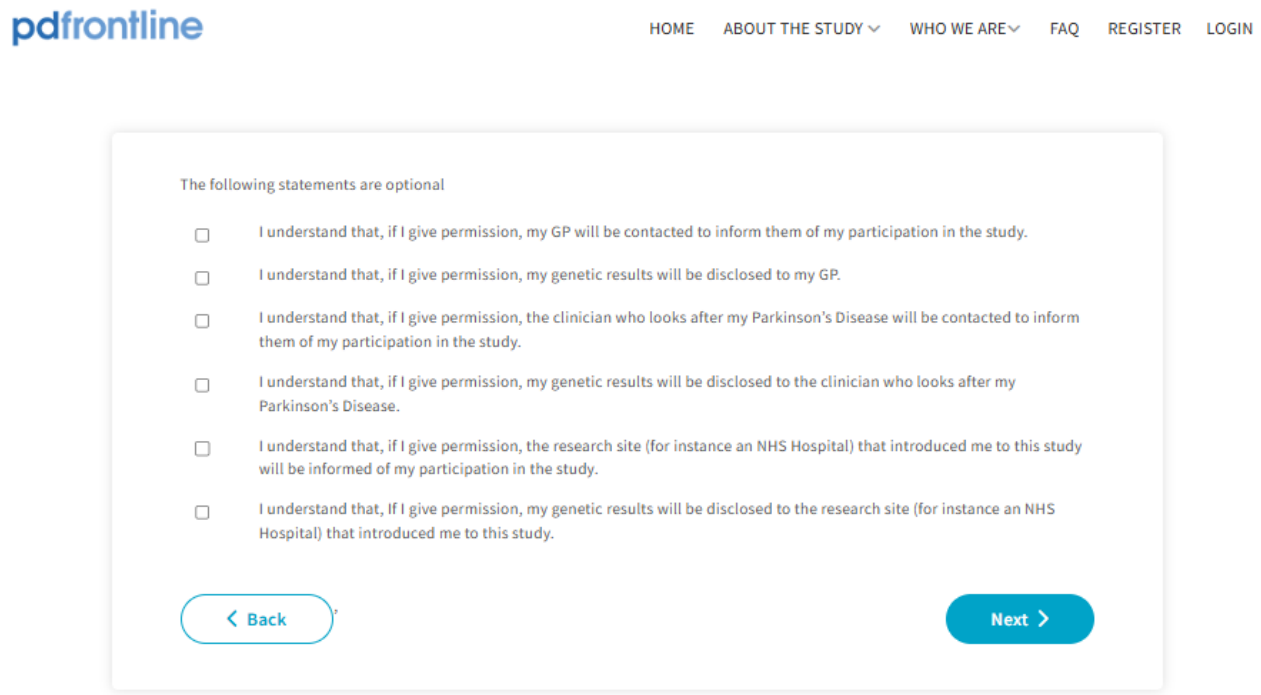
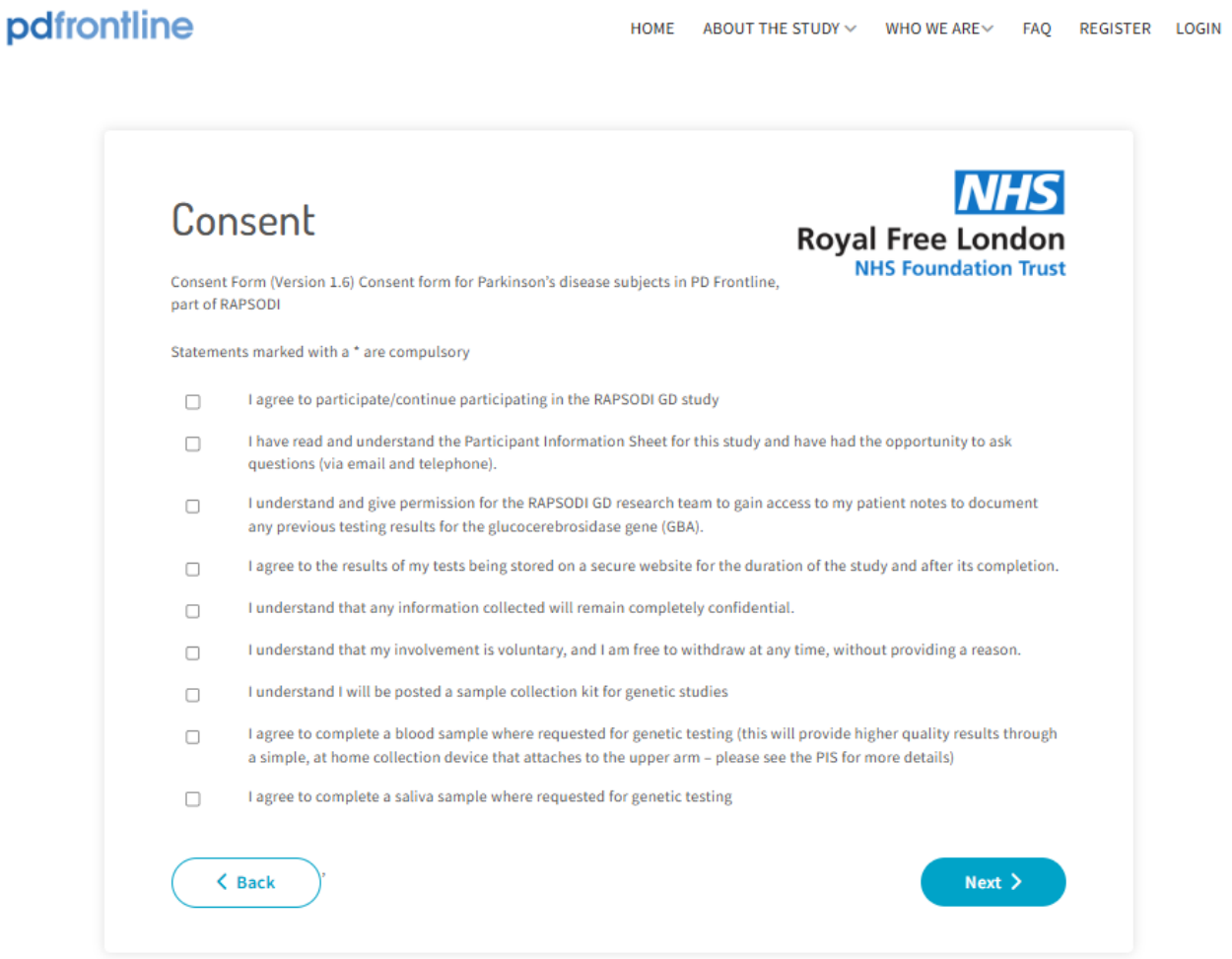
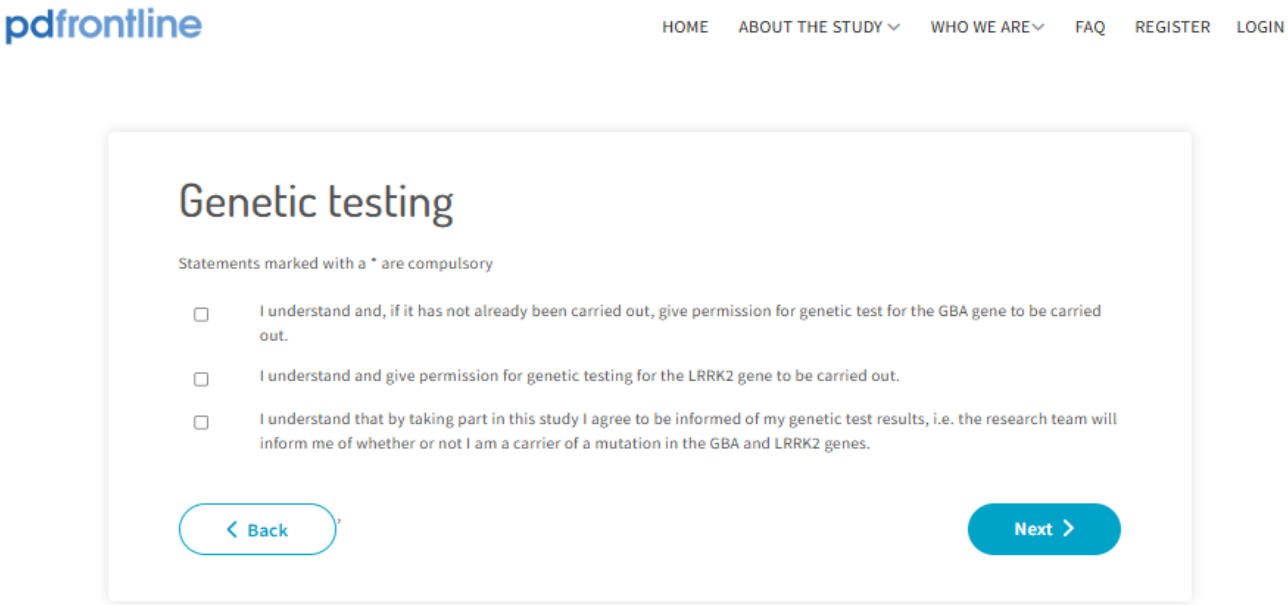
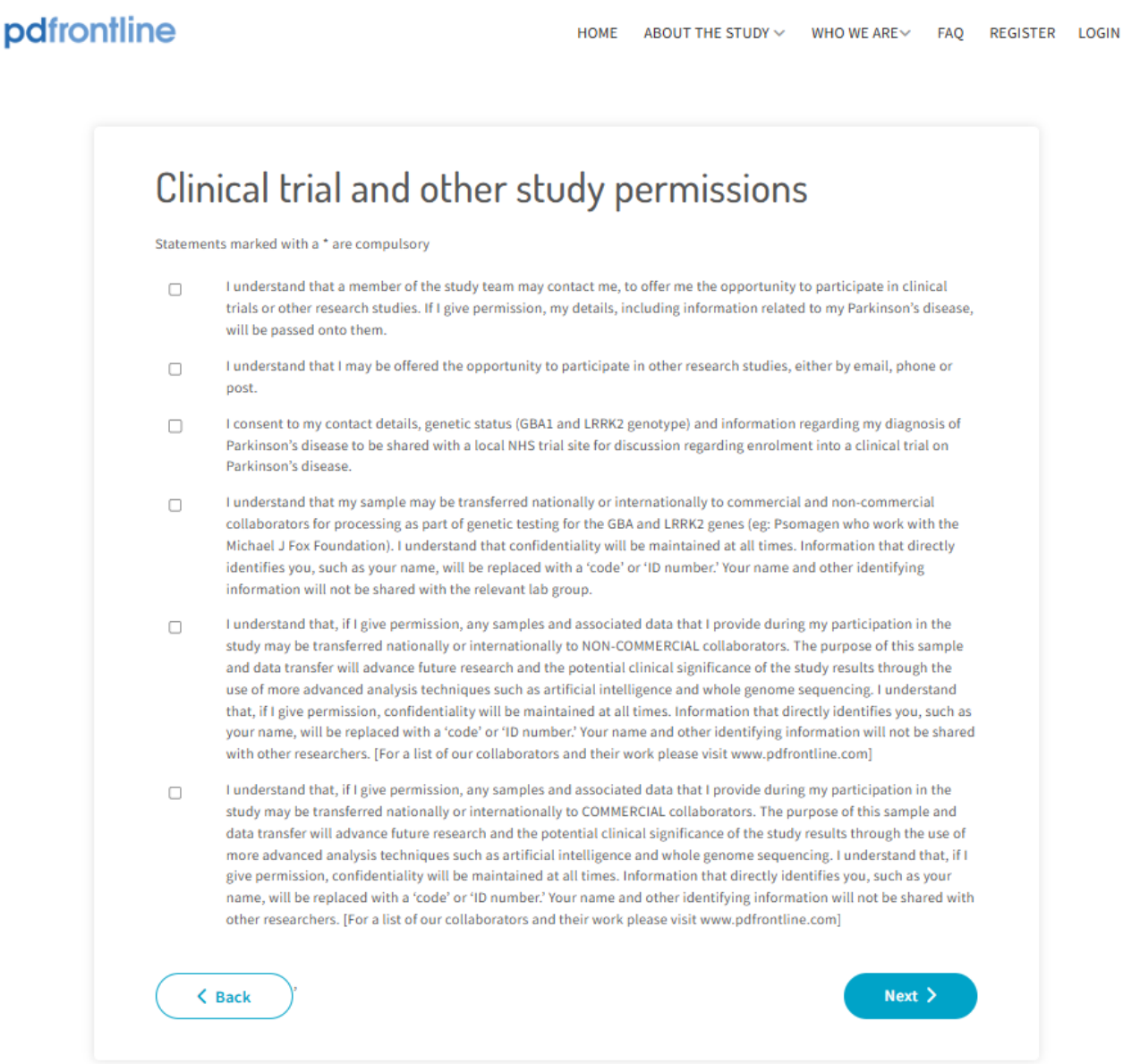


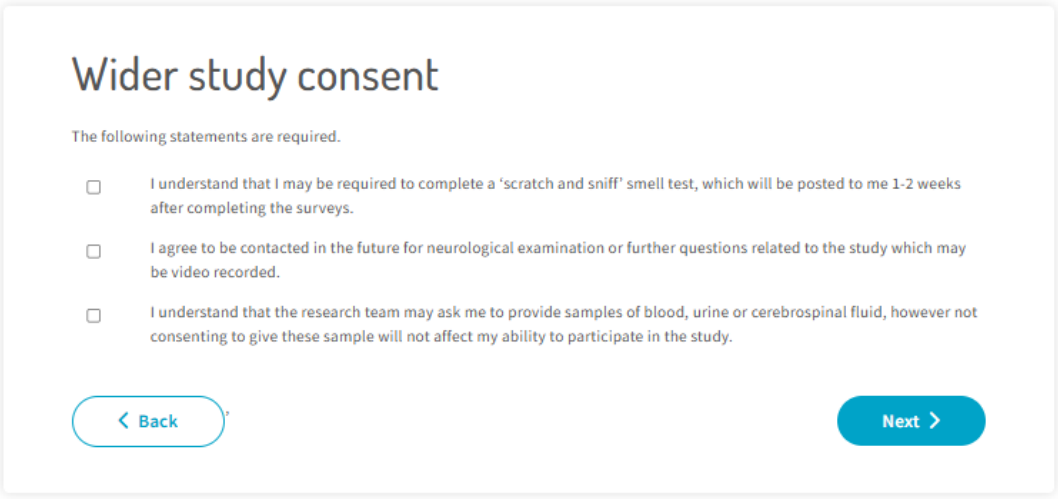
2. You will be taken to the Consent form as in the photo below. Please read each statement carefully and provide an answer for each. Questions that are compulsory to take part in the study are marked (*).



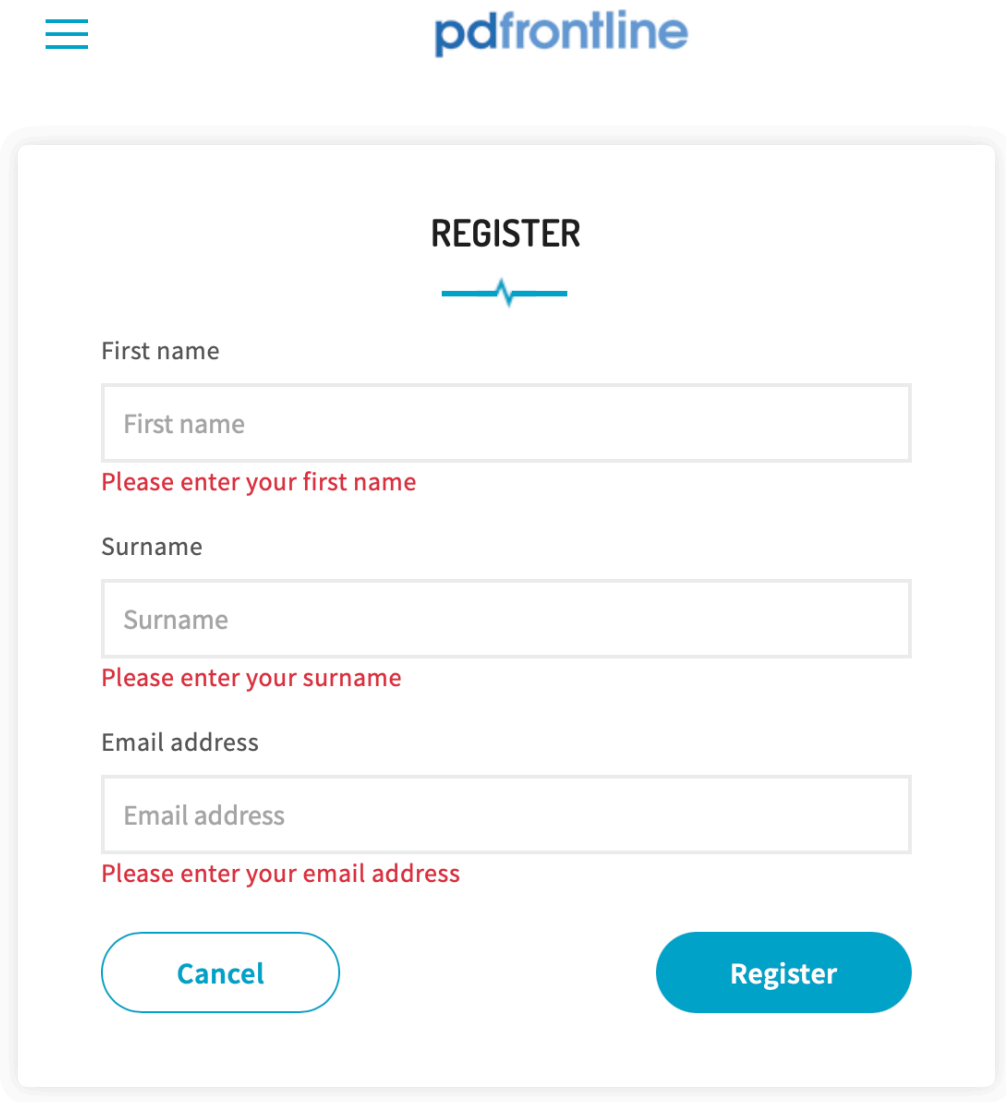




pdfrontline
HOME
ABOUT THE STUDY
WHO WE ARE
FAQ
REGISTER
LOGIN
Genetic testing
Statements marked with a * are compulsory
I understand and, if it has not already been carried out, give permission for genetic test for the GBA gene to be carried out.
I understand and give permission for genetic testing for the LRRK2 gene to be carried out.
I understand that by taking part in this study I agree to be informed of my genetic test results, i.e. the research team will inform me of whether or not I am a carrier of a mutation in the GBA and LRRK2 genes.
Back
Next

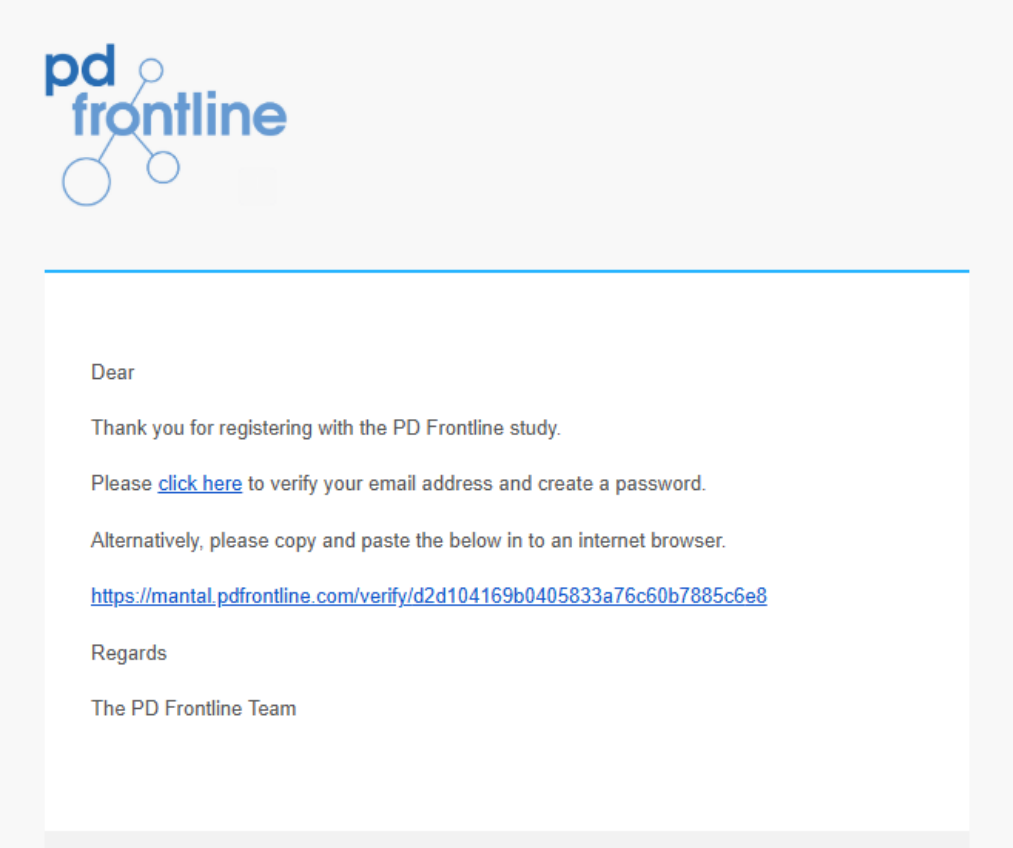

pdfrontline
HOME
ABOUT THE STUDY
WHO WE ARE
FAQ
REGISTER
LOGIN
Clinical trial and other study permissions
Statements marked with a * are compulsory
I understand that a member of the study team may contact me, to offer me the opportunity to participate in clinical trials or other research studies. If I give permission, my details, including information related to my Parkinson's disease, will be passed onto them.
I understand that I may be offered the opportunity to participate in other research studies, either by email, phone or post.
I consent to my contact details, genetic status (GBA1 and LRRK2 genotype) and information regarding my diagnosis of Parkinson's disease to be shared with a local NHS trial site for discussion regarding enrolment into a clinical trial on Parkinson's disease.
I understand that my sample may be transferred nationally or internationally to commercial and non-commercial collaborators for processing as part of genetic testing for the GBA and LRRK2 genes (eg: Psomagen who work with the Michael J Fox Foundation). I understand that confidentiality will be maintained at all times. Information that directly identifies you, such as your name, will be replaced with a 'code' or 'ID number.' Your name and other identifying information will not be shared with the relevant lab group.
I understand that, if I give permission, any samples and associated data that I provide during my participation in the study may be transferred nationally or internationally to NON-COMMERCIAL collaborators. The purpose of this sample and data transfer will advance future research and the potential clinical significance of the study results through the use of more advanced analysis techniques such as artificial intelligence and whole genome sequencing. I understand that, if I give permission, confidentiality will be maintained at all times. Information that directly identifies you, such as your name, will be replaced with a 'code' or 'ID number.' Your name and other identifying information will not be shared with other researchers. [For a list of our collaborators and their work please visit www.pdfrontline.com]
I understand that, if I give permission, any samples and associated data that I provide during my participation in the study may be transferred nationally or internationally to COMMERCIAL collaborators. The purpose of this sample and data transfer will advance future research and the potential clinical significance of the study results through the use of more advanced analysis techniques such as artificial intelligence and whole genome sequencing. I understand that, if I give permission, confidentiality will be maintained at all times. Information that directly identifies you, such as your name, will be replaced with a 'code' or 'ID number.' Your name and other identifying information will not be shared with other researchers. [For a list of our collaborators and their work please visit www.pdfrontline.com]
Back
Next



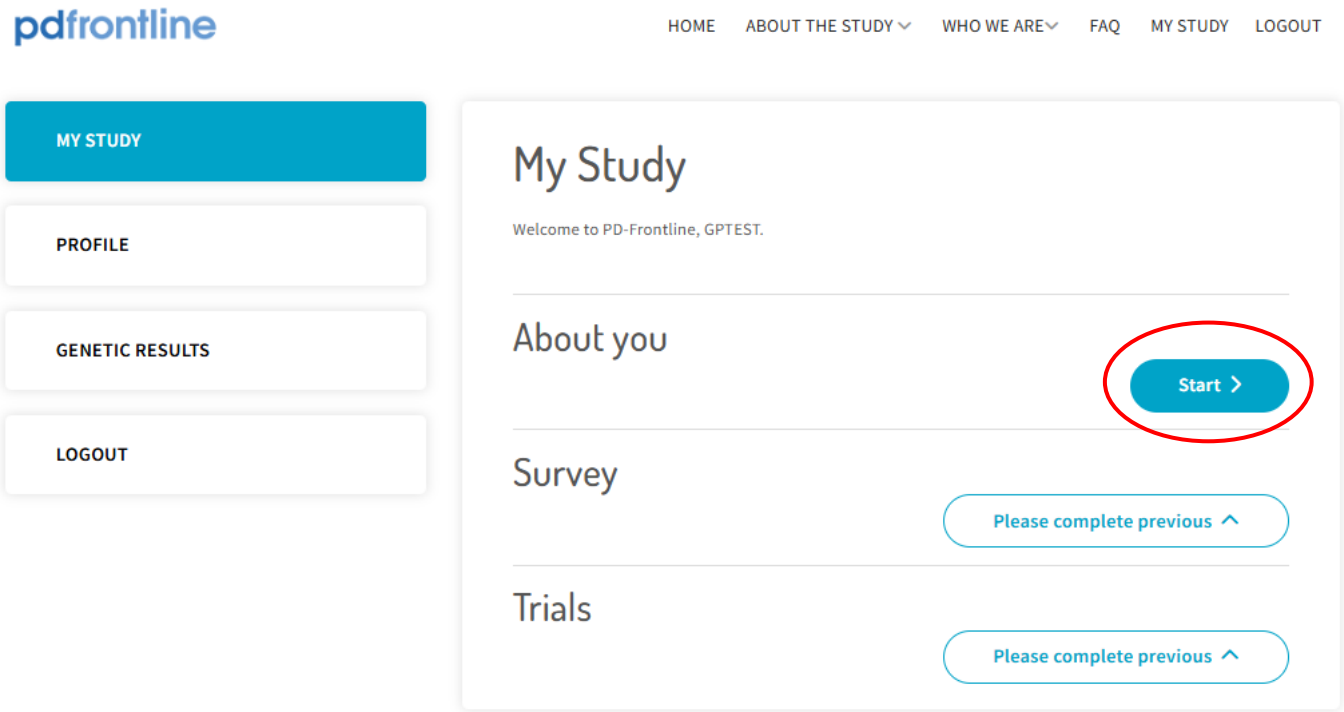

3. You will then be taken to the registration page where you will need to provide your name and an email address. This email address will be your username.



4. You will then receive an email with a verification link. Click the link to verify your account. The link will prompt you to create a password and then log you back into PD Frontline.




5. After logging in, you will then see your profile page as in the picture below; you can update your password and email preferences in the **'Profile'** tab. Your genetic results are available to download once ready from the **'Genetic Results'** tab. To start the study, you will need to click the blue **'Start'** button.

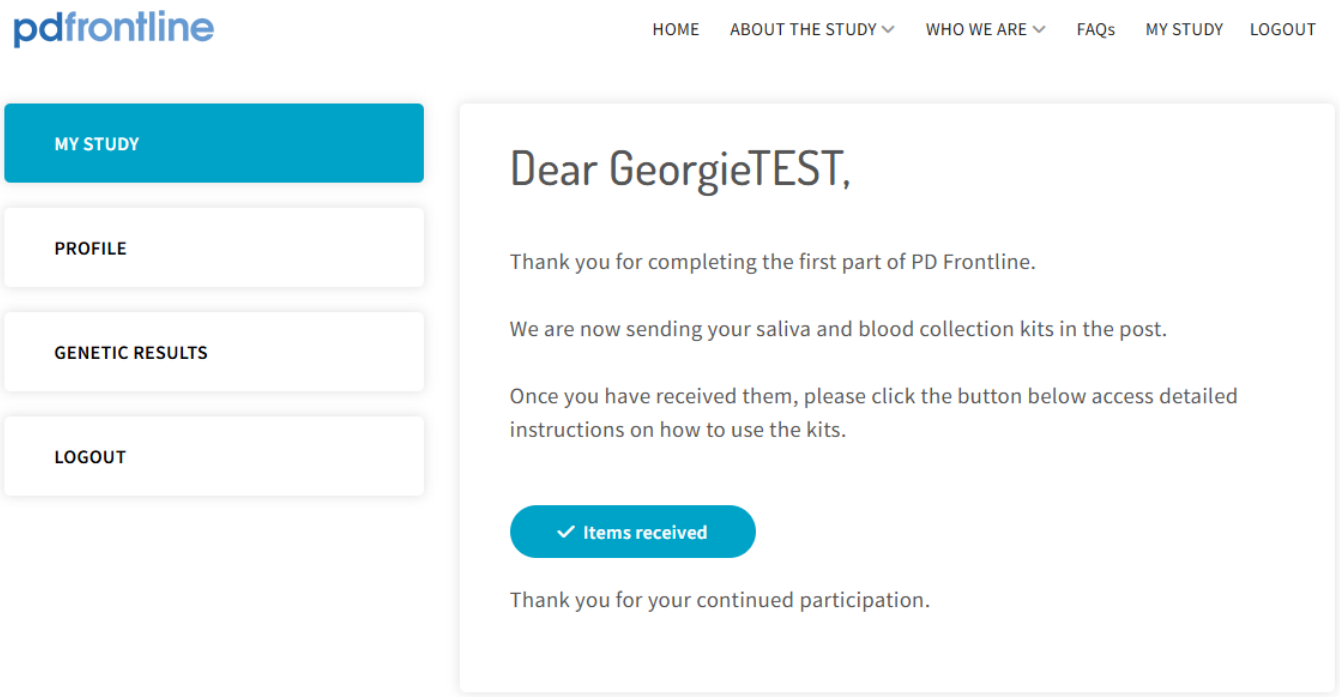


# Receiving a kit

After signing up, a DNA saliva and blood collection kit will be posted to you.

(Please allow up to three weeks for kits to arrive)

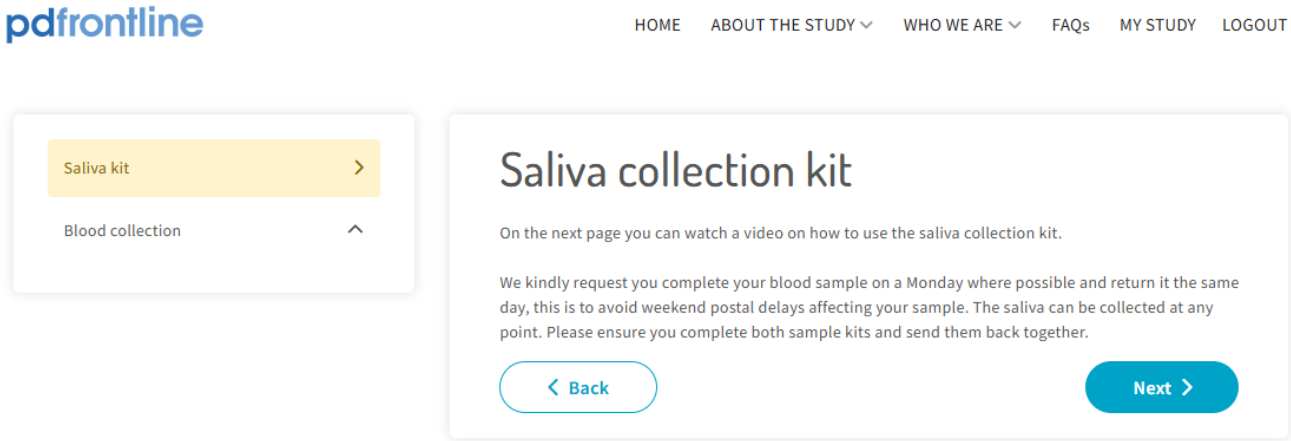
1. Once you receive your kit, please let us know by logging in and selecting the button: "**Items received**".

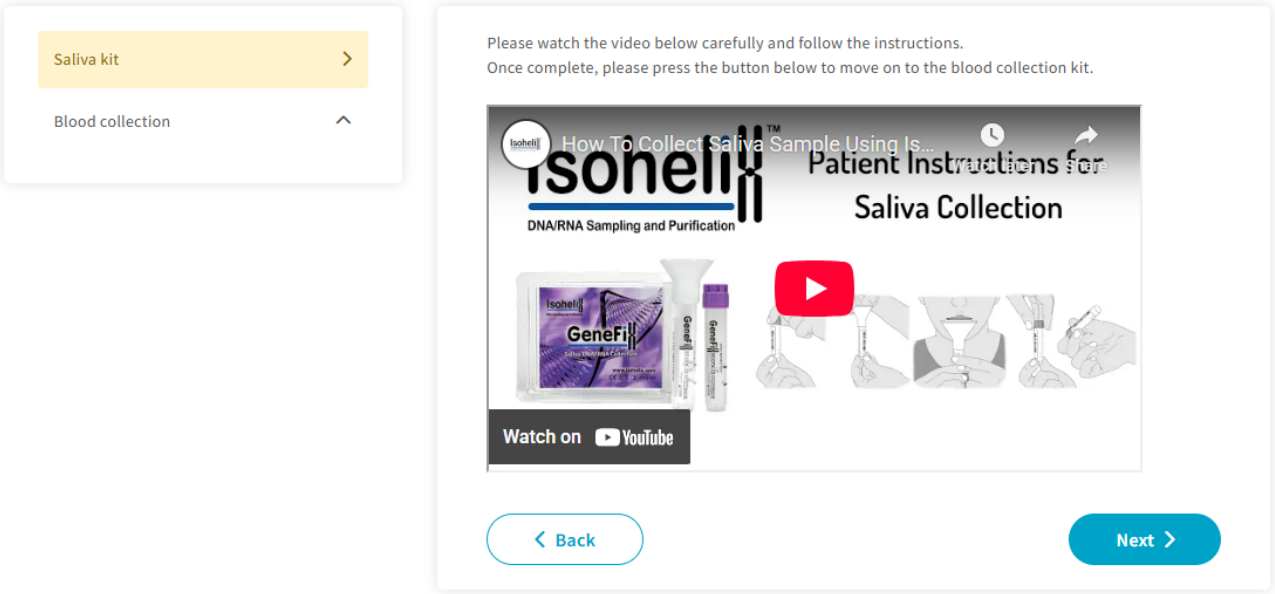



2. You will then be guided through instructional videos on how use the kits.

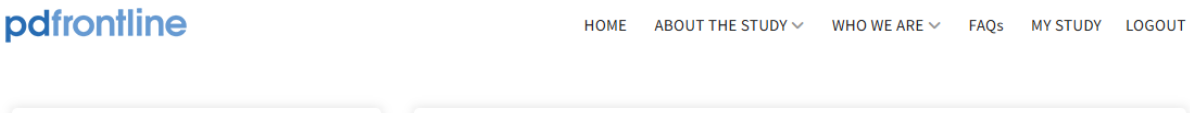
Please watch these videos carefully to ensure correct use. This helps us to obtain best quality DNA and reduces the chance of sequencing failure. **We kindly request you return your samples the day the blood is taken,** on a Monday where possible. This reduces the chance of your sample becoming unviable in the post.

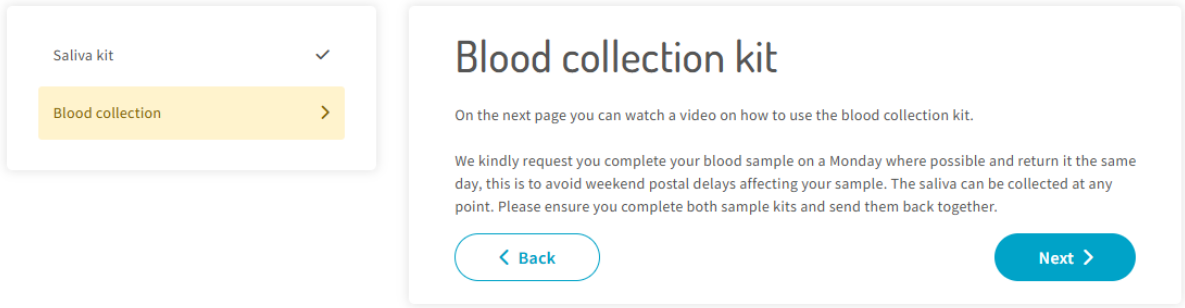
***Saliva kit***

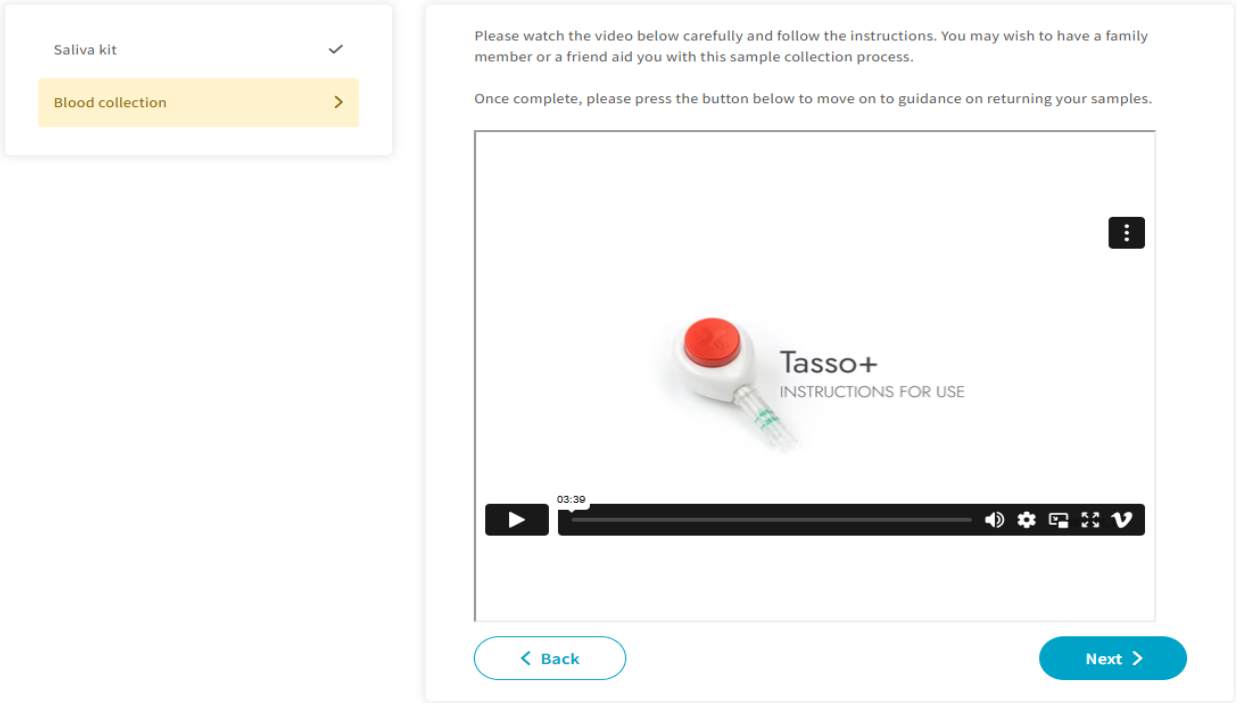





***Blood collection kit***



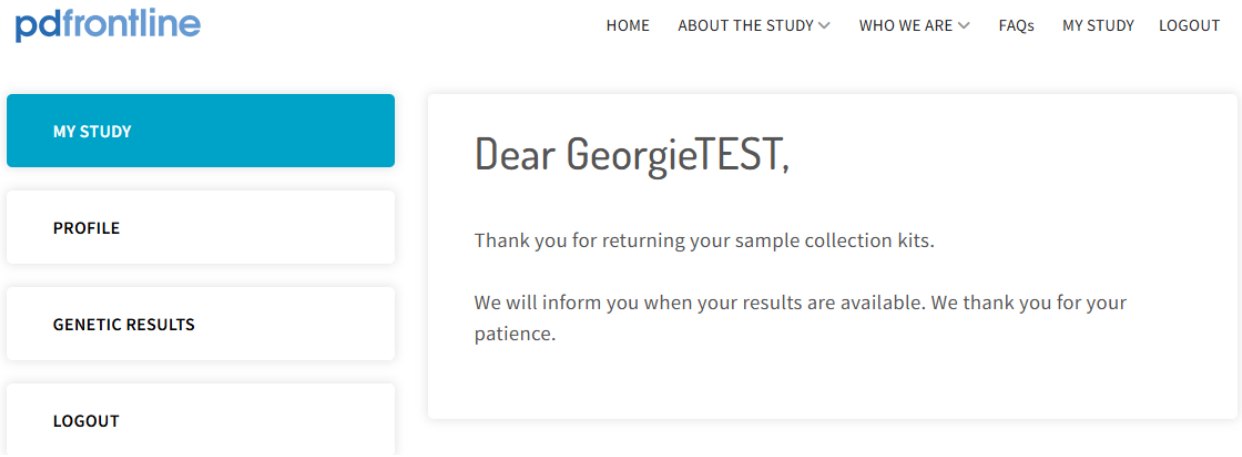



***Returning sample collection kits***

Please return both your blood and saliva sample using the prepaid white envelope provided.
**We kindly request you return your samples the day the blood is taken,** on a Monday where possible. This reduces the chance of your sample becoming unviable in the post.
We will notify you when we receive your samples and again when your results are available.

Additional instructional videos on how to pack and return the kits can be found on our YouTube channel: https://www.youtube.com/@pdfrontline

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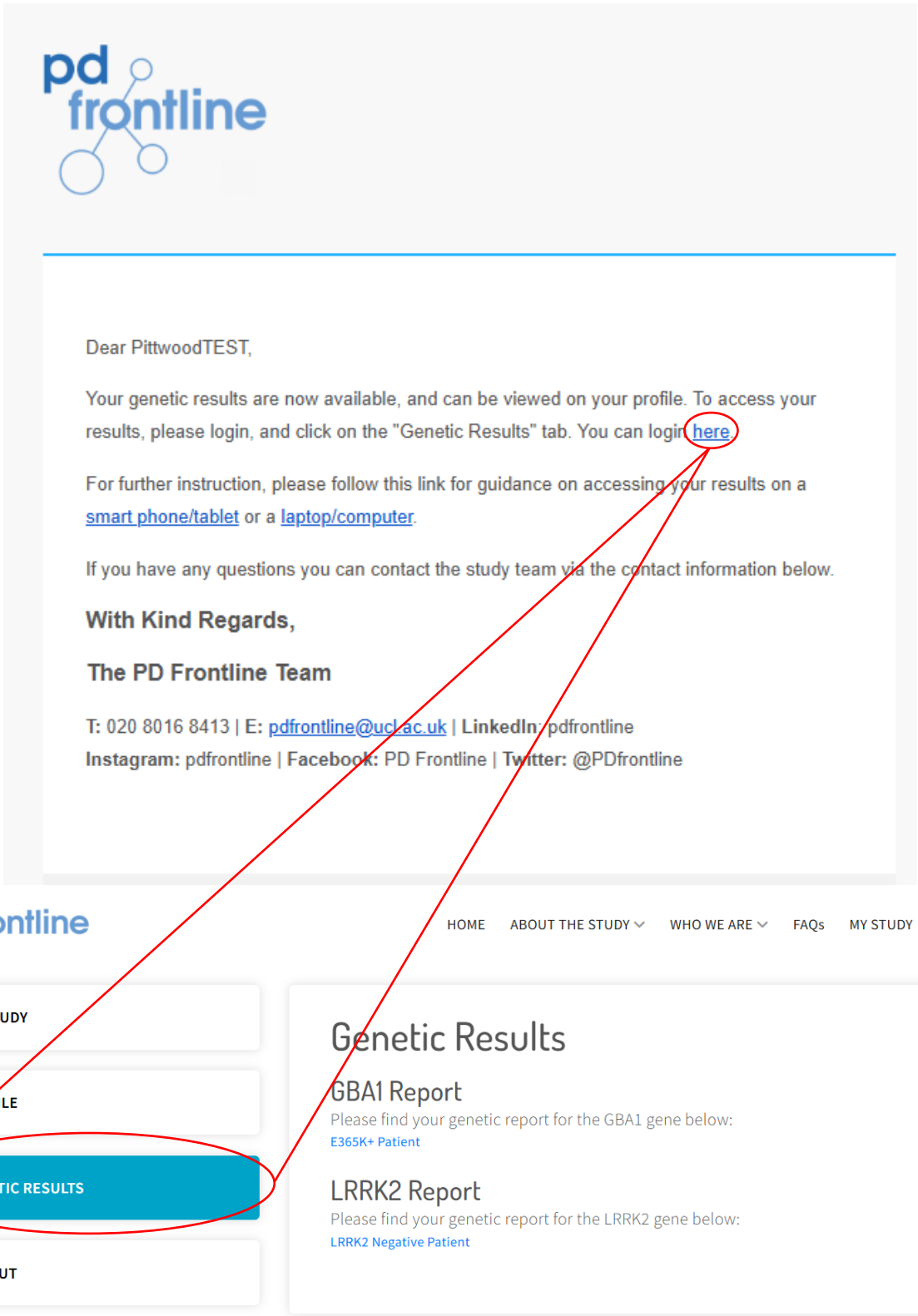
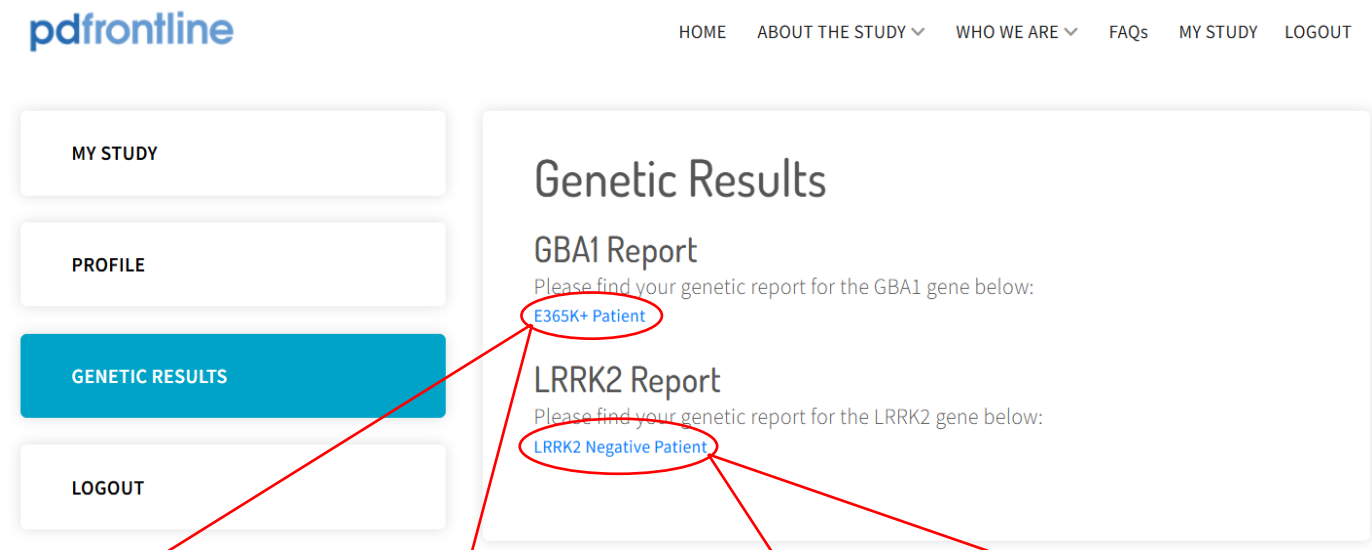
# Genetic results

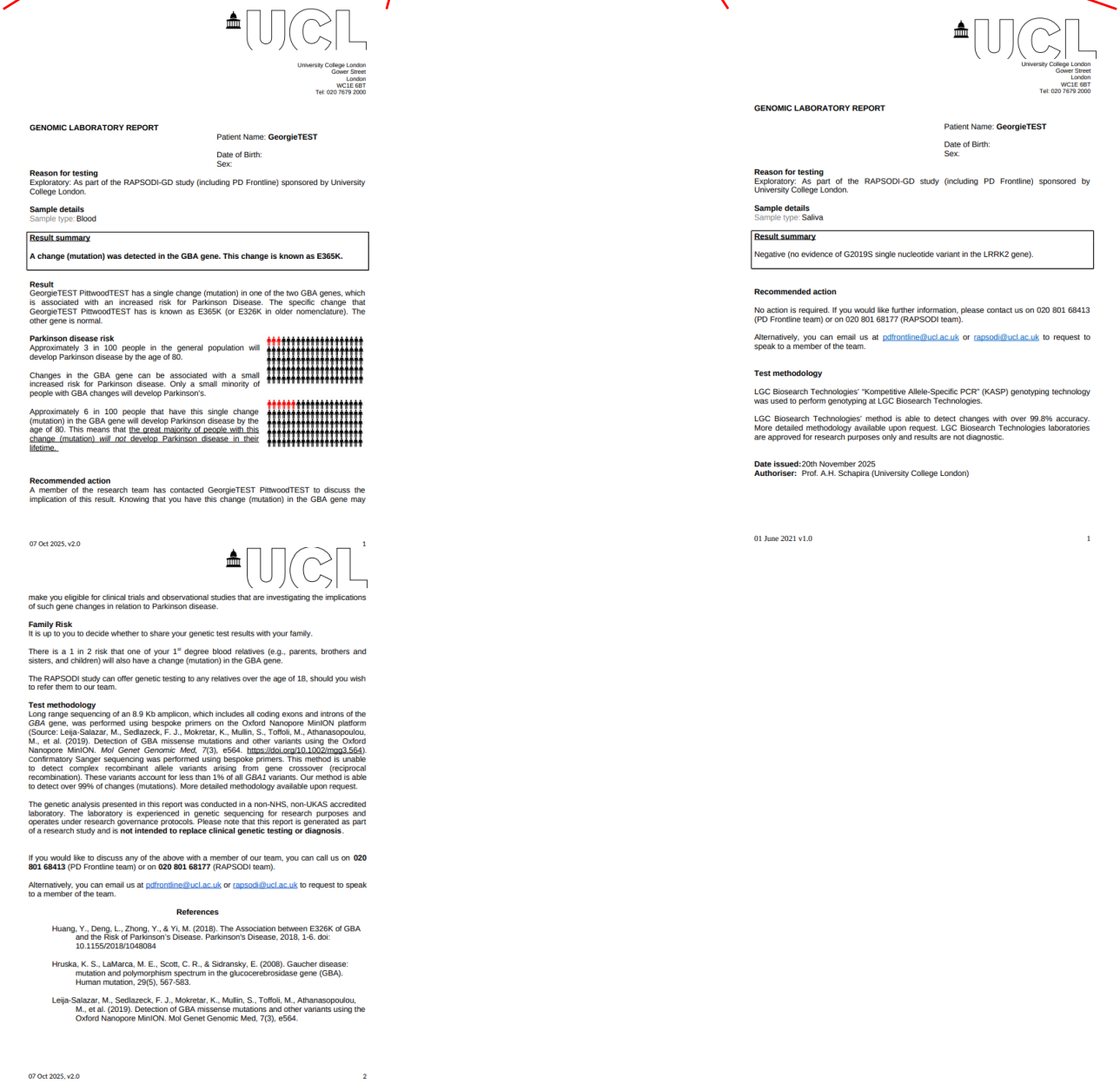
Your DNA samples will undergo genetic testing.

1. Once your genetic results are ready, you will receive an email or phone call to let you know.



2. To view your genetic results, click the **blue** link provided.
   This will download your genetic report as a PDF.



UCL
University College London
Gower Street
London
WC1E 6BT
Tel: 020 7679 2000

**GENOMIC LABORATORY REPORT**

Patient Name: **GeorgieTEST**
Date of Birth:
Sex:

**Reason for testing**
Exploratory: As part of the RAPSODI-GD study (including PD Frontline) sponsored by University College London.

**Sample details**
Sample type: Blood

**Result summary**

**A change (mutation) was detected in the GBA gene. This change is known as E365K.**

**Result**
GeorgieTEST PittwoodTEST has a single change (mutation) in one of the two GBA genes, which is associated with an increased risk for Parkinson Disease. The specific change that GeorgieTEST PittwoodTEST has is known as E365K (or E326K in older nomenclature). The other gene is normal.

**Parkinson disease risk**
Approximately 3 in 100 people in the general population will develop Parkinson disease by the age of 80.

Changes in the GBA gene can be associated with a small increased risk for Parkinson disease. Only a small minority of people with GBA changes will develop Parkinson's.

Approximately 6 in 100 people that have this single change (mutation) in the GBA gene will develop Parkinson disease by the age of 80. This means that the great majority of people with this change (mutation) *will not* develop Parkinson disease in their lifetime.

**Recommended action**
A member of the research team has contacted GeorgieTEST PittwoodTEST to discuss the implication of this result. Knowing that you have this change (mutation) in the GBA gene may

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make you eligible for clinical trials and observational studies that are investigating the implications of such gene changes in relation to Parkinson disease.

**Family Risk**
It is up to you to decide whether to share your genetic test results with your family.

There is a 1 in 2 risk that one of your 1st degree blood relatives (e.g., parents, brothers and sisters, and children) will also have a change (mutation) in the GBA gene.

The RAPSODI study can offer genetic testing to any relatives over the age of 18, should you wish to refer them to our team.

**Test methodology**
Long range sequencing of an 8.9 Kb amplicon, which includes all coding exons and introns of the *GBA* gene, was performed using bespoke primers on the Oxford Nanopore MinION platform (Source: Leija-Salazar, M., Sedlazeck, F. J., Mokretar, K., Mullin, S., Toffoli, M., Athanasopoulou, M., et al. (2019). Detection of GBA missense mutations and other variants using the Oxford Nanopore MinION. *Mol Genet Genomic Med*, *7*(3), e564. https://doi.org/10.1002/mgg3.564). Confirmatory Sanger sequencing was performed using bespoke primers. This method is unable to detect complex recombinant allele variants arising from gene crossover (reciprocal recombination). These variants account for less than 1% of all *GBA1* variants. Our method is able to detect over 99% of changes (mutations). More detailed methodology available upon request.

The genetic analysis presented in this report was conducted in a non-NHS, non-UKAS accredited laboratory. The laboratory is experienced in genetic sequencing for research purposes and operates under research governance protocols. Please note that this report is generated as part of a research study and is **not intended to replace clinical genetic testing or diagnosis**.

If you would like to discuss any of the above with a member of our team, you can call us on **020 801 68413** (PD Frontline team) or on **020 801 68177** (RAPSODI team).

Alternatively, you can email us at pdfrontline@ucl.ac.uk or rapsodi@ucl.ac.uk to request to speak to a member of the team.

**References**


Huang, Y., Deng, L., Zhong, Y., & Yi, M. (2018). The Association between E326K of GBA and the Risk of Parkinson's Disease. Parkinson's Disease, 2018, 1-6. doi: 10.1155/2018/1048084

Hruska, K. S., LaMarca, M. E., Scott, C. R., & Sidransky, E. (2008). Gaucher disease: mutation and polymorphism spectrum in the glucocerebrosidase gene (GBA). Human mutation, 29(5), 567-583.

Leija-Salazar, M., Sedlazeck, F. J., Mokretar, K., Mullin, S., Toffoli, M., Athanasopoulou, M., et al. (2019). Detection of GBA missense mutations and other variants using the Oxford Nanopore MinION. Mol Genet Genomic Med, 7(3), e564.


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University College London
Gower Street
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WC1E 6BT
Tel: 020 7679 2000

**GENOMIC LABORATORY REPORT**

Patient Name: **GeorgieTEST**
Date of Birth:
Sex:

**Reason for testing**
Exploratory: As part of the RAPSODI-GD study (including PD Frontline) sponsored by University College London.

**Sample details**
Sample type: Saliva

**Result summary**

Negative (no evidence of G2019S single nucleotide variant in the LRRK2 gene).

**Recommended action**

No action is required. If you would like further information, please contact us on 020 801 68413 (PD Frontline team) or on 020 801 68177 (RAPSODI team).

Alternatively, you can email us at pdfrontline@ucl.ac.uk or rapsodi@ucl.ac.uk to request to speak to a member of the team.

**Test methodology**

LGC Biosearch Technologies' "Kompetitive Allele-Specific PCR" (KASP) genotyping technology was used to perform genotyping at LGC Biosearch Technologies.

LGC Biosearch Technologies' method is able to detect changes with over 99.8% accuracy. More detailed methodology available upon request. LGC Biosearch Technologies laboratories are approved for research purposes only and results are not diagnostic.

**Date issued:** 20th November 2025
**Authoriser:** Prof. A.H. Schapira (University College London)

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