EXPRESS

Optimising EXome PREnatal Sequencing Services

July 2022

Parent & Family Newsletter

WELCOME...

...to the first edition of the EXPRESS newsletter for parents and families!

The national prenatal exome sequencing (pES) service launched in October 2020. This service is now offered as part of antenatal NHS care when scan findings suggest that a baby may have a genetic condition unlikely to be diagnosed by standard tests.

Because pES has not been offered in this way before, it is important that we evaluate how it is being delivered. This is the purpose of the EXPRESS study.

Over the next two years, our research team will evaluate the pES service. We will provide feedback about how it is being delivered across England so that we can ensure a service that both benefits and supports parents and families, and can be accessed equally by everyone.

The voices of parents and families are central to our research and the EXPRESS newsletter is where you can find out all you need to know about how we are involving parents and families in our work.

WHAT IS PRENATAL EXOME SEQUENCING?

Prenatal exome sequencing (pES) is a test that can be used to look for changes in an unborn baby's genetic code that may explain the unexpected features seen on an ultrasound scan.

This test is offered when other standard tests such as chromosomal microarray have not been able to find an explanation.

pES works by comparing an unborn baby's genetic code with both parents' (if possible) to help identify the difference between harmless changes that can run in families and changes that may be causing a genetic condition.

WHY IS PRENATAL EXOME SEQUENCING BEING OFFERED?

Until recently, pES had only been offered in a research setting. Now, this service is being delivered across the country as part of mainstream NHS care.

Because pES is able to look at genetic information in great detail, it can increase the chance of a genetic diagnosis, giving parents important information to help them identify treatment options or make decisions about their pregnancy.

What has EXPRESS done so far?

Finding out what healthcare professionals think of prenatal exome sequencing

Our research team has been interviewing healthcare professionals involved in offering pES, including fetal medicine and genetics doctors, genetic counsellors, and midwives, from every area of the country. In these interviews, we have asked how pES is being offered in their region, and the challenges and successes so far.

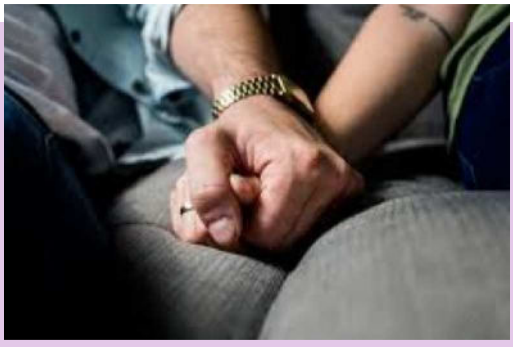
Finding out what online information is available to parents

We have researched the information available online, and have found that there is very little about exome sequencing that has been written with parents and families in mind (you can read more about what we found **here**).

As part of EXPRESS, we hope to develop more information resources and are currently creating an animation about pES with the input of parents and families, and healthcare professionals.

What's next for EXPRESS?

Parents' views and experiences of prenatal exome sequencing



A key part of EXPRESS is exploring parents' views and experiences of prenatal exome sequencing.

We have started to ask parents who have been offered exome sequencing in pregnancy (including parents who declined and parents who accepted the test) through fetal medicine units across the country and parent support organisations such as Antenatal Results and Choices (ARC).

We hope to gain an in-depth understanding of what it is like to be faced with unexpected news and the offer of genetic tests in pregnancy, and explore the information and support needs of parents in these circumstances. The ultimate aim is to make sure that exome sequencing is offered in the best possible way and parents receive appropriate support throughout the process.

How patients and the public help guide our research: Our Patient and Public Involvement Advisory Group (PPIAG)

Our research is guided by our **Patient and Public Involvement Advisory Group (PPIAG)**, who advise on study design, help construct parent information and suggest how to contact parents at this difficult time.

Our PPIAG is made up of patient advocates from a range of parent support charities and a researcher and parent who received a diagnosis in pregnancy. The PPIAG have been involved from the early stages in the design of the project and continue to give regular feedback to make sure the interests of parents and families are at the centre of our work.

Alström Syndrome UK
Strength for today, hope for the future

Breaking Down Barriers
Understanding genetics together

Genetic Alliance UK

Unique
Understanding Chromosome & Gene Disorders

SPOTLIGHT ON...

Dr Hannah McInnes-Dean

Hannah is a member of the EXPRESS team working at both Antenatal Results and Choices (ARC) and Great Ormond Street Hospital (GOSH). At ARC, she spends her time supporting parents who are making decisions before, during or after antenatal tests. At GOSH, she is a researcher working on the EXPRESS study where she is involved in interviewing healthcare professionals and parents to understand their views and experiences of prenatal exome sequencing.

We caught up with her to find out more about what Hannah's work involves.
Click here to watch a video of Hannah talking about how she mixes her research and supportive roles.

About the EXPRESS team

The EXPRESS team is a collaboration between clinicians, researchers and patient representatives from organisations across the UK. Considering parents' views is central to our work, so we are grateful to have Kerry Leeson-Beevers (Breaking Down Barriers) and Jane Fisher (ARC) to provide the parent voice to our research.

To find out more about who is involved, visit www.express-study.co.uk.

GET IN TOUCH!

If you want to find out more about the EXPRESS study or how you can get involved in our research, then we'd love to hear from you:

Email us at **express@nhs.gosh.uk**

Find out more about EXPRESS and the prenatal exome sequencing service at **www.express-study.co.uk**

Follow us on Twitter **@ES_EXPRESS**

If you would like information and support before, during and after antenatal screening, you can get in touch with Antenatal Results and Choices (ARC).

The helpline is answered by trained staff Monday to Friday, 10.00am-5.30pm.

You can also contact a member of the ARC team by email at **info@arc-uk.org**