mnda motor neurone disease association

Research

Inherited MND: Options when starting a family

Information Sheet B3

This publication gives information about the options available for those who have the rare inherited form of motor neurone disease (MND), and are wishing to start a family and do not want to pass on the risk of developing MND to their children. This information sheet has been written to explain the range of options available and their implications.

This information sheet is **part three** of our information on inherited MND.

- **Part one** of this information sheet looks at how motor neurone disease can be inherited and the importance of taking a full family history.
- **Part two** looks at genetic testing and how a diagnosis of inherited MND can affect insurance.

The content is split into the following sections:

Disclaimer: *Please note that information provided in this information sheet is based on a review of the currently available literature. This information sheet was written by the MND Association staff who are not clinicians and so any information provided in this sheet should not be considered as clinical advice. You should always discuss potential treatments with your clinician.*

This symbol is used to highlight **our other publications**. To find out how to access these, see *Further information* at the end of this sheet.

What do the words and abbreviations mean?

Amniocentesis	An invasive procedure using a needle to take a sample of the amniotic fluid.
Chorionic Villous Sampling (CVS)	An invasive procedure using a needle to take a sample of the placenta.
In Vitro Fertilisation (IVF)	A procedure during which an egg is fertilised by sperm outside of the body.
Pre-implantation Genetic Diagnosis (PGD)	A technique using an in-vitro fertilisation treatment to ensure that a faulty MND gene is not passed on to the baby.
Pre-natal Diagnosis (PND)	A procedure during pregnancy in which a sample is taken of the placenta or amniotic fluid to test the baby for an MND-causing gene.

1: Introduction

A small proportion (5-10%) of people with MND have a family history of the disease. This form of MND is known as familial, or inherited, MND. Inherited MND is caused by a mistake in the genetic code that holds the instructions for making every protein in our bodies; this mistake may be passed down from one generation to the next.

If you have a family history of MND (two or more 'blood relatives') or a family history of a type of dementia known as frontotemporal dementia (FTD) then you may be at risk of developing MND and passing the MND-causing gene on to your children in the future.

Whether or not an MND-causing gene has been identified in you or your family, there are several different options available for those wishing to start a family, which are explained in this information sheet.

We acknowledge that some of these choices may not be an option for everyone due to individual beliefs and ethical views. Therefore, it may be useful to talk over these options with a genetic counsellor.

I have inherited MND - why might I want to know which gene is causing it?

If you have a history of MND in your family, and are considering starting a family of your own, then you may be concerned about the risk that your child may develop MND in later life.

You may be feeling that the risk is too high, and that you would rather avoid having children. However, it is important to bear in mind that for many people MND does not

develop until later in life. In 40-50 years' time there may be a much greater prospect of curative treatments than there is today.

Is there any way to prevent MND being passed on to our baby?

The options open to you depend on your individual circumstances. For example, if you know which gene is causing MND in your family, it may be possible to test for it using pre-implantation genetic diagnosis (PGD) or pre-natal diagnosis (PND). These are tests which test for a particular gene defect (before or after conception) which has been identified as causing MND in your family.

If you do not know the gene defect which is causing MND in your family, but wish to have children, you may want to consider gamete (sperm or egg) donation or adoption. These options mean that the gene defect is not passed on to the child, but the child may not be fully biologically yours.

Each of these options will be discussed individually in this information sheet, providing information in order for you to consider which may be the best option for you. For example, you may not wish to know your carrier status (whether you carry the gene defect) so some options may not be appropriate. Age may be a factor in your decision, along with the costs and time frames.

2: Pre-implantation Genetic Diagnosis (PGD)

If you know which gene defect is causing MND in your family, it may be possible to undergo a technique called pre-implantation genetic diagnosis (PGD).

PGD is a technology that uses In Vitro Fertilisation (IVF) treatment to ensure that the faulty gene causing MND in your family is not passed on to the baby. IVF is performed to create embryos. These embryos are then tested at around 5 - 6 days of development when they are made up of a small number of cells (about the width of a human hair in size). The testing can tell which embryos are affected and unaffected by MND. Any embryos free of the MND-causing gene can then be transferred to the mother's womb with the hope that she will become pregnant and have a healthy baby.

Is PGD possible for all couples who are at risk of passing on MND to their child?

PGD will not be an option for everybody. For PGD to take place the faulty gene that is causing MND in your family must be known (SOD1, TDP-43, FUS, C9orf72). This gives rise to a number of issues; for example, if the MND is caused by a faulty gene that is yet to be identified then it will not be possible to develop a test, and therefore PGD will not be possible.

PGD can be a lengthy process (around 9 - 12 months before the start of treatment), and for this reason there are age restrictions for those wishing to go ahead. In all cases couples may be eligible for treatment on the NHS if, at the time of being referred for PGD, the prospective mother is under 40 years old. This age limit is set because the success of PGD decreases significantly with age. However, in some cases, the age limit may be lower if it is a new form of MND that has not been tested before (e.g., a new gene). This is because it takes longer to prepare PGD for such couples.

For each first case of this nature an application for a licence needs to be approved by the Human Fertilisation and Embryology Authority (HFEA) before PGD can take place. Furthermore, once a licence has been issued, a new test needs to be developed which is specific to that gene.

Licences are currently approved for MND caused by mutations to the SOD1 or C9ORF72 gene.

Will I need to find out my carrier status?

The implications of finding out for sure whether or not you have inherited the faulty gene are significant, and for this reason many people choose not to do so. Most people going through PGD have had a test and know that they have the MND causing-gene. This type of PGD is called '**direct testing PGD**'.

However, in some instances, it may be possible for you to go ahead with PGD without finding out whether you have inherited the faulty gene, provided that other family members have been tested for the faulty MND gene and been found to carry it. This is called '**exclusion PGD**'

If your parent has MND and has been tested, then, provided the gene defect has been identified, PGD is theoretically possible. If there are cases of MND in your family and, if you or your 'at risk' partner does not wish to know their carrier status, PGD may be possible by means of 'exclusion testing'. This is where your carrier status can remain unknown as long as you know the faulty gene that is causing MND in your family. This is currently being offered for Huntingdon's disease and it may be possible for people with MND.

It is important to know that if you choose not to know your carrier status you may be undergoing PGD treatment unnecessarily.

First steps

The first step will be to speak to your GP, who will make referrals to your local genetic service. It is important to note that some PGD centres only accept referrals from genetic centres. Local funding bodies:

England: NHS Commissioning Board and Clinical Commissioning Groups (previously known as Primary Care Trusts)
Wales: Welsh Health Services Specialist Commissioning
N. Ireland: Health and Social Care Boards

For further information and to download an information sheet on PGD, please see the Genetic Alliance UK and HFEA resource (find the link in the 'How do I find out more?' section).

3: Pre-natal Diagnosis (PND)

If you know which gene defect is causing MND in your family, it may be possible to undergo a process called pre-natal diagnosis (PND).

PND involves an invasive diagnostic procedure that can be performed after 10 weeks of pregnancy. A sample is taken from either 11-14 weeks (chorionic villous sampling) or between 15 -20 weeks (amniocentesis) to test the baby for the MND-causing gene. Because these are invasive procedures, they carry a risk of miscarriage. The risk is between 1 in 100 and 1 in 200.

Chorionic villous sampling (CVS) is a procedure that uses a needle through the abdomen (or very occasionally the cervix) to take a tiny sample of placental tissue.

Amniocentesis is another invasive test that uses a needle to take a tiny sample of the amniotic fluid surrounding the baby. You may be more familiar with these procedures in the context of testing for Down syndrome.

If the faulty gene is present, then it is possible to detect this at an early stage of pregnancy and you may then decide to undergo termination of pregnancy. If the gene is not present, then you can continue with the pregnancy knowing that your baby will be free from the gene causing MND in your family.

Is PND possible for all couples who are at risk of passing on MND to their child?

PND will not be an option for everybody for a variety of reasons. Like PGD, for PND to take place the gene defect that is causing MND in your family must be known. If the MND is caused by a faulty gene that is yet to be identified, then it will not be possible to develop a test for it, and therefore PND will not be possible.

It is best to speak with a genetic centre about testing before you become pregnant, so you can make sure a test (and PND) would be available. PND, unlike PGD, is not a lengthy process, and for this reason there are no age restrictions for those wishing to go ahead with it. As the test is done after conception has occurred, the only requirement is that the faulty gene in the family has been identified in order for the foetus to be tested.

It is important to know that PND for late onset adult disorders (like MND), for which there is no preventative treatment, is done only in cases where couples are sure that they would not continue the pregnancy if the faulty gene was to be identified. This is because continuing with the pregnancy breaches the child's right to choose whether or not to be tested.

Will I need to find out my carrier status?

Your local genetic centre would test the 'at risk' parent for the faulty gene beforehand if PND testing was requested. This would mean that you would know whether or not you carry the faulty gene.

However, like PGD, it may be possible for you to go ahead with PND without finding out whether you have inherited the faulty gene by means of exclusion testing, provided that other family members have been tested for the faulty MND gene and been found to carry it. This means your carrier status can remain unknown as long as you know the gene defect that is causing MND in your family. It is important to know that if you choose not to know your carrier status you may be undergoing PND treatment unnecessarily.

How much does PND cost?

PND testing by amniocentesis or CVS is available on the NHS without any special need to apply for funding. PND testing is usually arranged through your regional genetic service.

How long will it take to get the results after PND?

The results from PND should take around 2 - 3 weeks to come through.

First steps

The first step will be to speak to your GP, who will make referrals to your local genetics service.

For further information and to download an information leaflet on PND, please see the Genetic Alliance UK and Antenatal Results and Choices resource (find the link in the 'How do I find out more?' section).

4: Gamete donation

If you do not know which gene is responsible for causing MND in your family, or do not want to risk passing on the MND-causing gene to your child, you may wish to consider gamete donation.

Gamete donation is where you can create a baby using an egg or sperm donation. If a woman has a family history of MND then a donated egg is fertilised using her partner's sperm using IVF, as described earlier. If a man has a family history of MND then donated sperm is used to fertilise his partner's egg by artificial insemination.

Is gamete donation possible for all couples who are at risk of passing on MND to their child?

Gamete donation will not be an option for everybody; however, it is an option if the faulty gene in your family has not yet been identified through research.

If a woman has a family history of MND then gamete donation can be a lengthy process due to IVF, and for this reason there are age restrictions for those wishing to go ahead. In all cases, couples may be eligible for treatment on the NHS if, at the time of being referred for IVF, the prospective mother is under 40 years old. This age limit is set because the success of IVF decreases drastically with age.

If a man has a family history of MND then gamete donation can be a much simpler process. Using ultrasound, the donated sperm is inserted into the womb when the woman is most fertile (sometimes fertility drugs are used to increase egg production). This is known as intrauterine insemination (IUI).

You may choose not to consider gamete donation as an option as the child will not be biologically yours (the parent who is at risk of developing MND), which may be unacceptable for some couples.

How much does gamete donation cost?

If a woman decides to undergo gamete donation, if you fit the criteria, you may be eligible for three cycles of IVF on the NHS and you will need to make an application for funding to your local funding body. The chances of securing funding can depend on where you live and your local funding body's criteria for funding. Self-funding is an option and the cost will be in the region of £5,000 per cycle, but could be more depending on the clinic you use.

If a man decides to undergo gamete donation, if you fit the criteria, you may be eligible for six cycles of artificial insemination on the NHS and you will need to make an application for funding to your local funding body. Self-funding is an option and the cost will be in the region of £500 to £1,000 per cycle.

Couples should be aware that pregnancy success with IVF and artificial insemination is not guaranteed.

The HFEA website has a list of fertility clinics along with their success rates. If you are investigating clinics for treatment you should ask if they are accredited centres, how many cycles they have done, and what their accuracy rate and success of treatment are.

Will I need to find out my carrier status?

No. If preferred, prospective parents can go ahead with gamete donation without knowing their carrier status as the gene defect does not need to be known prior to gamete donation.

Should I tell my child about their origins?

Evidence from experiences of adoption and studies of donor-conceived people suggests that it is best to tell people about their origins in childhood as finding out later in life may be emotionally damaging. Therefore, being open with your child from an early age is important. It is advisable to explore this issue thoroughly before proceeding so that you have a good understanding of the implications of creating a family through gamete donation.

If you have received treatment at an HFEA licensed clinic, you can contact the HFEA to find out information about the child's donor.

First steps

The first step will be to speak to your GP and local fertility clinic. Before treatment is offered, it is essential that you speak to a genetic counsellor.

To find out more information about genetic counselling, see Information Sheet B2 - *Genetic testing and insurance*.

For further information on gamete donation please see the Donor Conception Network and HFEA resource (find the link in the 'How do I find out more?' section).

5: Adoption

If you do not know which gene defect is responsible for causing MND in your family, or if you do not fit the criteria/cannot afford the previous options or find them unacceptable, then you may prefer to consider adoption.

Adoption is a legal procedure in which all the parental responsibility is transferred to the individual or couple who have adopted the child. An adopted child loses all legal ties with their biological mother and father (the 'birth parents') and extended family, and becomes a full member of the new family.

Is adoption possible for all individuals who are at risk of passing on MND to their child?

Adoption will not be an option for everybody; however, it is an alternative if you do not want to undergo genetic testing or do not know the faulty gene causing MND in your family.

Adoption is a service designed to find loving and secure families for children who, for a variety of reasons, are unable to be cared for by their birth families. When considering adoption it is important to learn more about the process and the children who need a new family through adoption. Information can be obtained from the agencies listed at the end of the information sheet.

Adoption agencies hold information and preparation sessions where these things can be explored. Following an application to adopt, a full assessment is carried out on all applicants to determine their suitability to adopt, and this includes a comprehensive health assessment.

The assessment process typically takes at least 6 months, while the social workers get to know you and understand what you have to offer as parents. You must be over the age of 21 and there is no upper age limit. At the end of the assessment period, an adoption panel will hear all about you and make a recommendation to the agency about your suitability to adopt.

The **Coram BAAF Adoption and Fostering Academy** state that although "health issues will need to be explored prior to adoption, health problems and disabilities are not barriers to adoption, provided you can care for the child you adopt".

Each applicant is considered on an individual basis, which involves completion of a health form and a comprehensive assessment by their GP, with a report sent to the adoption agency's medical adviser. Further information may be requested from relevant consultants with your consent, for example a clinical geneticist, in order to assess current and potential future risks to your health. Any concerns about your health will need to be explored with your social worker, to see how you might manage different possible scenarios and how these might impact on parenting. If you have had experience of MND in your family then this may give you added strengths that the adoption agency and panel will also consider, such as coping with grief, care, life experiences, commitment and flexibility in responding to needs.

During the assessment process you will also have opportunities to consider the age and characteristics of the child/children you are best suited to parent. It is important to provide information to the adoption agency so that they understand that a family

history of MND (in which a gene defect has been identified or not) only increases your risk of developing MND, and you may or may not develop it in the future.

It is also important to know that when applying as a couple, if there are concerns about the health of the 'at-risk' partner then the adoption panel will take into consideration the health and motivation of the other partner before making a decision.

Children who are being placed for adoption will have experienced loss, and may have experienced inadequate parenting, neglect and/or abuse so you would need to be aware of this and consider how best to meet the needs of that individual child.

It may also take a long time in order for the right child to become available, particularly if you want to adopt a younger child or a baby.

We appreciate that some couples may not choose adoption because the child will not be biologically theirs.

How much does adoption cost?

Adoption is free, although some agencies expect applicants to pay for the required health assessment. As the fee for this is set by their GP, it can be up to £200. In some cases, the adoption agency will cover the cost of the fee. Applicants will need to undergo a comprehensive assessment and be approved by an adoption agency in order to adopt.

Will I need to find out my carrier status?

No, if preferred you can go ahead with applying for adoption without knowing your carrier status. If the gene causing MND in your family has been identified, you may need to discuss your reasons for not wanting a genetic test with your social worker. If the gene causing MND in your family has not yet been identified, then you will be unable to find out your carrier status.

How do I tell my child they are adopted?

This can be a challenging topic but being open with your child from an early age is important. Coram BAAF has produced a booklet called 'Talking about adoption' which aims to prepare and support parents when telling their child about their origins.

First steps

The first step will be to speak to your local adoption agency, which can be found using this website: **www.corambaaf.org.uk/agencies**. For more detailed information about the adoption process in your part of the UK, the following organisations can help:

England: www.first4adoption.org.uk
Wales: www.adoptcymru.com
N. Ireland: www.adoptionandfostering.hscni.net
Scotland: www.scottishadoption.org

For further information on adoption, see the Coram BAAF resource (find the link in the 'How do I find out more?' section).

6: Comparison table

The choices available for couples when starting a family, which are listed in this information sheet, are compared in the table below so that you can quickly see which options may be best suitable for you.

For example, if the prospective mother is over the age of 40 then PGD may not be an option, but PND and adoption will be.

If you do not know which gene is causing MND in your family, as it has not yet been identified through research, or you have not undergone genetic testing, then PGD and PND will not be an option, but gamete donation and adoption will be.

	PGD	PND	Gamete donation	Adoption
Will I need to know which gene is causing MND in my family?	Yes	Yes	No	No
Will I need to know my carrier status?	Yes*	Yes*	No	No+
How much does it cost (private treatment)?**	~£10,000	Free	~£5,000 per MF cycle; £800-£1,300 for insemination	Up to £200 health assessment fee
Is there an age limit?	<40 years (mother)	No	<40 years (mother) for IVF	>21 years
Will my child be biologically mine?	Yes	Yes	No	No
How long does it take?	Can be lengthy, depends on funding	Up to 20 minutes for procedure + 2-3 weeks for results	Can be lengthy, depends on funding	>6 months

* It may be possible to go ahead with PGD/PND without knowing your carrier status by means of exclusion testing.

** Correct time at publishing

+ If the gene causing MND in your family has not been identified through research, then you will not know your carrier status. If the gene has been identified, then you may need to know your carrier status when undergoing a full health assessment.

7: Care and support

The care required by someone with inherited MND is no different to a person who has the non-inherited form of the disease, and the MND Association has a range of services available to help. You can speak to your GP, local fertility clinic or adoption agency regarding the choices available when starting a family. Referral to a neurologist who understands the particular needs of people with inherited MND may be helpful.

I would like to know more about genetic testing

If you have MND and would like to know which gene is responsible for causing the disease in your family you may be able to have a genetic test. This will allow you to know if you have a disease-causing gene that has been shown by research to cause inherited MND (SOD1, TDP-43, FUS and C9ORF72). There are still a number of genes which have not yet been identified, so testing for these will not be available at present.

Once a gene defect has been identified your unaffected family members may wish to be tested to see if they too have the gene responsible for causing MND in your family.

I would like to know more about genetic testing

A family history of inherited MND may affect life and travel insurance, which may be something that worries you and your family. It is important to know that there is a great deal of information and support available regarding this topic.

For more information on inherited MND, see Information sheet: B1 - *Introduction to inherited motor neurone disease*.

For more information about insurance, see Information sheet: B2 - *Genetic testing and insurance*.

8: How do I find out more?

Useful organisations

We do not necessarily endorse any of the following organisations but have included them to help you begin your search for further information.

The contact details are correct at the time of publishing but may change between revisions. If you need help to find an organisation, contact the Research Development Team (see *Further information* at the end of this sheet for details).

Genetic Alliance

The Genetic Alliance UK is an organisation that aims to improve the lives of people affected by a genetic condition. They have a number of leaflets and documents under the 'Information Centre' tab on their website, including one on insurance.

Address: 4D Leroy House, 436 Essex Road, London, N1 3QP
Email: contactus@geneticalliance.org.uk
Telephone: 0207 704 3141
Website: **www.geneticalliance.org.uk**

British Society for Genetic Medicine

Has a directory of UK regional genetic centres so that you can find your local centre.

Address: Charles Darwin House, Roger Street, London, WC1N 2JU
Email: membership@bsgm.org.uk
Telephone: 0203 793 7851
Website: **www.bsgm.org.uk**

The Centre for Pre-implantation Genetic Diagnosis

Has general information about PGD including conditions tested and information about referrals and funding.

Address: 11th Floor, Tower Wing, Guy's Hospital, Great Maze Road, London, SE1 9RT
Email: PGDGenetics@gstt.nhs.uk
Telephone: 0207 188 1364
Website: **www.pgd.org.uk**

Human Fertilisation and Embryology Authority (HFEA)

Has a large amount of information on PGD, IVF and gamete donation, as well as a list of the genes PGD is currently approved for testing, as well as those awaiting consideration. They can also provide details for local HFEA approved fertility clinics.

Address: 10 Spring Gardens, London, SW1A 2BU
Email: enquiriesteam@hfea.gov.uk
Telephone: 0207 291 8200
Website: **www.hfea.gov.uk**

Antenatal Results and Choices (ARC)

Provides non-directive information and support around antenatal testing and its consequences.

Address: 345 City Road, London, EC1V 1LR
Email: info@arc-uk.org
Telephone: 0845 077 2290 or 0207 713 7486 from a mobile
Website: **www.arc-uk.org**

CoramBAAF Adoption and Fostering Academy

Provides information about adoption and fostering throughout the UK, and the criteria and assessment procedure. They produce a range of information booklets and can provide you with details of your local adoption agency.

Address: Coram Campus, 41 Brunswick Square, London, WC1N 1AZ
Email: advice@corambaaf.org.uk
Telephone: 0207 520 0300 or Advice Line 0300 222 5775 (Mon-Fri, 9am-1pm)
Website: **www.corambaaf.org.uk**

The Donor Conception Network

Aims to support families through donor conception. They have information and personal stories of donor conception and produce a number of information booklets.

Address: 154 Caledonian Road, London, N1 9RD
Email: enquiries@dcnetwork.org
Telephone: 0207 278 2608
Website: **www.dcnetwork.org**

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Further information

You may find these information sheets from the MND Association helpful:

B1 – *Introduction to inherited MND*

B2 – Inherited MND: Genetic testing and insurance

We also provide the following guides:

Living with motor neurone disease – our main guide to help you manage the impact of the disease

Caring and MND: support for you – comprehensive information for unpaid or family carers, who support someone living with MND

Caring and MND: quick guide – the summary version of our information for carers

You can download most of our publications from our website at **www.mndassociation.org/publications** or order in print from the MND Connect helpline, who can provide further information and support.

MND Connect can also help locate external services and providers, and introduce you to our available services, including your local branch, group, Association visitor or regional care development adviser.

MND Connect
Telephone: 0808 802 6262
Email: mndconnect@mndassociation.org
MND Association, David Niven House, 10-15 Notre Dame Mews, Northampton NN1 2BG

Research Development Team
Telephone: 01604 611 880
Email: research@mndassociation.org

MND Association website and online forum
Website: **www.mndassociation.org**
Online forum: **forum.mndassociation.org** or through the website

We welcome your views

Your feedback is really important to us, as it helps improve our information for the benefit of people living with MND and those who care for them. If you would like to provide feedback on any of our information sheets, you can access an online form at: **www.surveymonkey.co.uk/r/infosheets_research**

You can request a paper version of the form or provide direct feedback by email: research@mndassociation.org.

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