Pregnancy and reproduction

Where there is a family history of a muscle disorder both men and women may have a number of concerns when they are considering having children. The potential problems may be different depending on the type of muscle disorder and the sex of the parent. In addition to the concerns outlined in this leaflet there may be more particular genetic issues which should be discussed with the Genetic Specialists in your own locality.

What effect does being pregnant have on muscular dystrophy?
The extra weight associated with being pregnant is generally the main problem for women who have weakness, especially if they are at the stage where they have difficult walking and frequently trip or fall. Occasionally women may find that they need to use a wheelchair most of the time at the end stage of their pregnancy and this may further aggravate their weakness. Returning to full strength following delivery and the reduction of their weight may be difficult. In addition both men and women will need to consider the problems associated with lifting, carrying and nursing their baby. Remember though that none of these difficulties will be unique, and that someone may well have thought of solutions to them. There are lots of ideas about how to deal with these kinds of issues on the Internet.

Thinking about difficulties associated with specific conditions, women with myotonic dystrophy are at risk of a number of complications which include increased risk of miscarriage, increased fluid around the baby (hydramnios) which may cause premature labour, heavy bleeding either before or following delivery and retained placenta or afterbirth. Many of these complications are particularly evident when the baby has congenital myotonic dystrophy. The incidence of diabetes is slightly increased in myotonic dystrophy and the bodily changes associated with pregnancy may precipitate diabetes during pregnancy, which will bring its own complications.

Occasionally the mother’s muscle weakness may make it harder for the mother to push with contractions, and this may result in the baby needing help at delivery, either by forceps or ventouse (suction applied to babies head). This is generally done under local anaesthetic. If the baby is very distressed or the mother is a long time in labour, the mother may have an emergency caesarean section. If problems are anticipated this may be the planned form of delivery.

Caesarian section is either performed under epidural (when the mother is awake) or under general anaesthetic (GA). This is a major operation and it may take additional time for women with muscle weakness to recover their strength fully. Particular care needs to be taken for patients with myotonic dystrophy having GA, as they have an increased risk of complications with the use of certain drugs. All patients with muscular dystrophy are at increased risk of developing chest infection following GA, this risk is increased for smokers.

In individuals who have weak breathing muscles or major curvature of the spine (scoliosis), the growing pregnant abdomen can reduce breathing capacity further, so a full respiratory assessment is advisable pre pregnancy. This includes measurement of lung volumes, and if these are below about 50% predicted, monitoring breathing overnight (sleep study). Should these tests show significant problems, use of a small breathing machine (non invasive ventilation) during sleep can maintain oxygen carbon dioxide levels, and may allow a successful pregnancy, in some case.

Those with heart involvement from their muscular dystrophy may find that pregnancy precipitates or brings on additional problems. Pregnancy causes the body to increase the amount of fluid. This, together with the extra weight from the developing baby may increase the work the heart needs to perform. Delivery may put additional strain on the heart. If you have concerns about this type of
problem then you should discuss them with your doctors. Not all patients with muscular dystrophy will be at risk of heart complications.

What about the genetic implications?
There are different forms of inheritance in muscular dystrophy. The confirmation of your precise diagnosis may help the doctors advise you as to the risks of your baby having the same problem as you. Sometimes the form of inheritance may be worked out from the pattern of inheritance in your family. There may be variation in the severity of affected individuals between families and within families. Myotonic dystrophy tends to get more severe when it is passed from parent to child. This increase is more marked when a mother passes on the condition.

Males with sex or X-linked conditions such as Duchenne and Becker will not have affected sons. Their daughters will all be carriers and are at risk of having affected sons.

If the condition is genetic what are the options for prenatal diagnosis?
Gene tests are now possible for many types of muscular dystrophy. You need to discuss this with your doctor to find out exactly what could be available for you. Some families may need to have the specific form of the alteration identified within their family. If a specific test is available then you may opt for a test on the pregnancy to see if the baby is affected or not. This may not be entirely straightforward as tests may not always be able to predict how severe the baby’s problems will be, so you really need to talk through the issues relevant to your own particular case.

What kinds of tests are available?

Chorionic Villus Sampling/Biopsy (CVS/CVB)
This test involves removing a small portion of the placenta or afterbirth from within the womb by inserting a needle through the abdominal wall just below the belly button or umbilicus (occasionally this test is done via the vagina). The needle goes through the muscles of the womb and is guided using an ultrasound scan. The procedure is performed from 11-12 weeks of pregnancy and takes about 20 minutes. Only specialised centres perform this test. The miscarriage risk is about 1 in 50. The test results are generally available within 2 weeks. Some tests including sexing for sex or x-linked conditions may be available within a couple of days. It is important that you check details of the testing including the miscarriage risk with your local centre.

Amniocentesis
This is performed in a similar fashion to a CVS, but it involves the removal of a small amount of fluid from around the developing baby within the womb. Most main maternity units perform the test. It is performed from 15 weeks with a miscarriage risk of about 1 in 100. The results take about 3 weeks.

Termination
You may choose to have prenatal testing with the intention of terminating an affected pregnancy. Before 12 weeks this may be performed under general anaesthetic by sucking the pregnancy from the womb. More frequently all terminations are now induced medically with drugs used to induce the opening of the cervix (opening of the womb). After 12 weeks this is the normal method and the drugs are used to induce an early labour and natural delivery. This is to ensure that the womb and cervix are not damaged to reduce the complication for future pregnancy and the fertility. Occasionally not all the contents of the womb, particularly the afterbirth are expelled naturally, and the woman may need an operation under general anaesthetic to remove the retained products. If you have opted for a termination or are thinking of doing so then you may wish to contact the organisation ARC (Antenatal Results and Choices) on 020 7631 0285 or web: www.arc-uk.org. ARC offers non-judgmental support and further information.

Scans for sex determination
For sex linked conditions these scans may be performed from about 20 weeks. Sexing the baby depends on the development of the external genitals of a male. Occasionally these may be underformed and the sex may not be accurate at this stage.
Preimplantation Genetic Diagnosis (PGD)
At present, only a few centres in the UK perform this procedure. For each patient a special license must be obtained from the Human Fertilisation and Embryology Authority. It combines in vitro fertilisation (IVF) or test tube pregnancy plus genetic testing.

The woman is given drugs to over stimulate the ovaries to produce lots of eggs. These are then harvested by inserting a tube near the ovary via the vagina. The eggs are put in a dish and fertilised with the father’s sperm. They are grown until there are about eight cells. One cell is then carefully removed and tested genetically. Eggs unaffected by the specific form of muscular dystrophy, which are developing normally, are selected to be inserted into the woman’s womb. Generally two eggs if available are inserted to increase the chance of pregnancy. Drugs are then used to help ensure that the womb is receptive for the developing eggs. The potent drugs used may cause side effects. About four out of every ten women who have IVF take home a baby following the procedure. Those who are successful may have a multiple or twin pregnancy.

This may be an option for women who have fertility problems and have an identified gene alteration associated with a high risk of having an affected child. The unit may advise having either a CVS or an amniocentesis to confirm the test. There may not be any suitable eggs produced or the only eggs may have the gene alteration. Some units may store unused eggs or embryos for future pregnancy attempts. If these are not stored or required they need to be destroyed.

Egg/Sperm Donation
Women or men with a condition may wish to avoid having a child with their particular condition by asking for a donation of eggs or sperm from a donor woman or man. Sperm is easier to donate than eggs. Egg donation generally comes from women who are having IVF and have spare eggs. It involves stimulation of the women with particular drugs. Sperm donation is relatively straightforward and involves insertion of the donated sperm into the womb at the time of ovulation via a tube in the vagina. The resulting child will not be at risk of the relevant condition as they will not be the genetic offspring of the relevant parent. New legislation means that the children have the right to access the identity of the genetic or natural parents following egg or sperm (gamete) donation.

Adoption/Surrogate Mother
These are other options that are beyond the scope of this leaflet. Details may be obtained via specialised agencies.

What about infertility?
This may occur for a number of reasons. Some patients may have difficulty having sexual intercourse due to physical disability such as spinal deformity or contracture.

Muscle weakness may cause physical difficulty in assuming the appropriate position, or maintaining the physical exertion required for the sexual act. These types of problems may result in incomplete penetration or maintenance of the sexual act. Those making love infrequently may not perform the act at the time when the female’s eggs are available for fertilisation. If the woman has a regular monthly menstrual cycle of 28 days (while not on the oral contraceptive pill) ovulation occurs 14 days after the start of the period, and fertilisation may occur following intercourse about 2-3 days before or after this date. Males with myotonic dystrophy may have reduced fertility. This is associated with changes in the testes, but the exact causes of these are not yet known.

Of course, infertility is potentially a problem for anyone, and couples may have trouble conceiving for unrelated reasons. Most couples who have regular unprotected intercourse will have conceived within one year, if this is not the case then you should consult with your GP about referral to a special infertility unit. Funding for such treatment is varied across the country and may not be available for couples who already have a child.

Is impotence a problem with muscular dystrophy?
Erectile dysfunction or impotence (difficulty for the male to develop or maintain a hard penis) is not a problem caused by muscular dystrophy. It may be due to a number of other problems. It is common in
diabetics, which may in some cases be associated with neurological problems. Psychological issues that may be related to physical disability or dislike of self-image, can often cause major problems. In women this may show as difficulty relaxing and enabling or enjoying sexual intercourse. Help may be obtained via your GP, local Family Planning Clinics, or via the Relate (Marriage Guidance Counselling Service) who employ specialist sexual health advisors.

Viagra (Sildenafil) may be helpful for the treatment of some patients, but can be contra-indicated in patients with heart problems, which are of course commonly associated with some forms of muscular dystrophy. So, while Viagra may be helpful for some patients, it is definitely something that should be discussed with your doctor.

Parenting
Pregnancy is not the end of the problem. In many ways it may only be the start! When you have considered all these options remember that if you are successful in having a child, that the child will need to be cared for and nurtured for many years. This may have both physical and psychological implications, which may weaken or strengthen your ability to cope with your own disability. As with all these issues, it is very important to talk this through with your partner and family, and with the doctors and support staff who know you, and can help you come to an informed decision.

Disabled Parents Network (DPN) is a National organisation of and for disabled people who are parents or who are to become parents and their families, friends and supporters.

Helpline: 08702 410 450
Web: www.disabledparentsnetwork.org.uk

Related factsheets produced by the Muscular Dystrophy Campaign
- Inheritance and the muscular dystrophies.
- Organisations that provides information, advice and support on issues surrounding personal relationships and sexuality.

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