Genetic Advice and Support Workshop
Action Duchenne 2014

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Workshop

- Your experiences and questions
- Information sheet development
- Things we could discuss
  - Genetic Counselling
  - DMD and BMD
  - Carrier tests
  - Inheritance of DMD and BMD
  - Pregnancy options
  - Carrier health
  - Talking about DMD in the family
Genetic Counselling

Offered by local Genetics Services
See a medical doctor or a genetic counsellor
Establish a diagnosis
Draw a family tree
Discuss how DMD/BMD is inherited
Figure out who needs advice
Testing of the DMD gene in affected boys or potential carriers
Put families in touch with specialist services
Describe DMD to relatives at risk
Discuss family planning options
Duchenne Muscular Dystrophy
DMD

- Affects about 1 in 3000 boys
- Progressive muscle weakness
- Female carriers may be a mother, sister, aunt or niece of an affected boy
- Carriers don’t usually have significant muscle problems
- Carriers can have boys with or without DMD, girls who are carriers or not carriers
- A milder form of DMD is called Becker MD
Male - 46,XY

DMD gene
X and Y

DMD Gene is called dystrophin
Female - 46,XX

DMD gene
The DMD gene

- One of the biggest genes known
- Can be altered in many different ways
- Makes a protein important for muscle structure and function
X-linked Recessive Inheritance

Sperm: X Y

Female carrier: X X

Eggs:
- 25% chance Non-carrier female
- 25% Carrier female
- 25% Healthy male
- 25% Male with DMD
Who can be a carrier: Family trees

- Male
- Female
- Carrier
- DMD
- DMD
- DMD

DMD
No family history of DMD

About 2 out of 3 times Mum will be a Carrier

About 1 out of 3 times Mum will not be a carrier
What does Genetic testing do?

- Confirms diagnosis
- Localises the exact gene alteration
- Provides a test for relatives especially potential carriers
- Might influence therapy
- Allows reproductive choice
Carrier tests

- **CK** (creatine kinase) test
  - Blood test
  - Not always reliable

- **Genetic test**
  - Looks at the DMD gene
  - If the ‘spelling mistake’ in the family is already known then carrier testing is accurate
Carrier Health Issues

Do I need health checks?

- Female carriers may be at increased risk of cardiomyopathy (heart disease) but the extent of this is debated.
- Some doctors may recommend heart screening but this has been questioned.
- A small % of carriers may have skeletal muscle problems.
- Be sensible about symptoms.
Having Children

- Whether or not to have any tests to see if a pregnancy is affected by DMD
- Testing at birth (CK or gene test)
- Finding out if it's male or female
- Having a prenatal gene test
- Having IVF to select an embryo (PGD) not affected by DMD
Pregnancy

Female carriers of DMD can have Free Fetal DNA sexing from around 8 weeks

male

female
Pregnancy

Female carriers of DMD can be offered prenatal diagnosis

Chorionic villus sampling (CVS) around 12 weeks
Amniocentesis 16 weeks

1-2% miscarriage risks
Preimplantation Genetic Diagnosis: PGD

sperm → egg

Test single cells from each embryo

Healthy embryo

mother

~8X

~20% success
Genetic Counselling

[Genetic family tree diagram]

You
Example 1

DMD

carrier

Am I a carrier?
Example 2

DMD

Am I a carrier?

Not a carrier
Mosaicism

BLOOD

EGGS
Support Groups and Information

- Action Duchenne
  - www.actionduchenne.org

- Muscular dystrophy Campaign
  - www.muscular-dystrophy.org

- Contact a Family
  - www.cafamily.org.uk

- Muscular Dystrophy Association
  - Mda.org

- Antenatal results and Choices
  - www.arc-uk.org