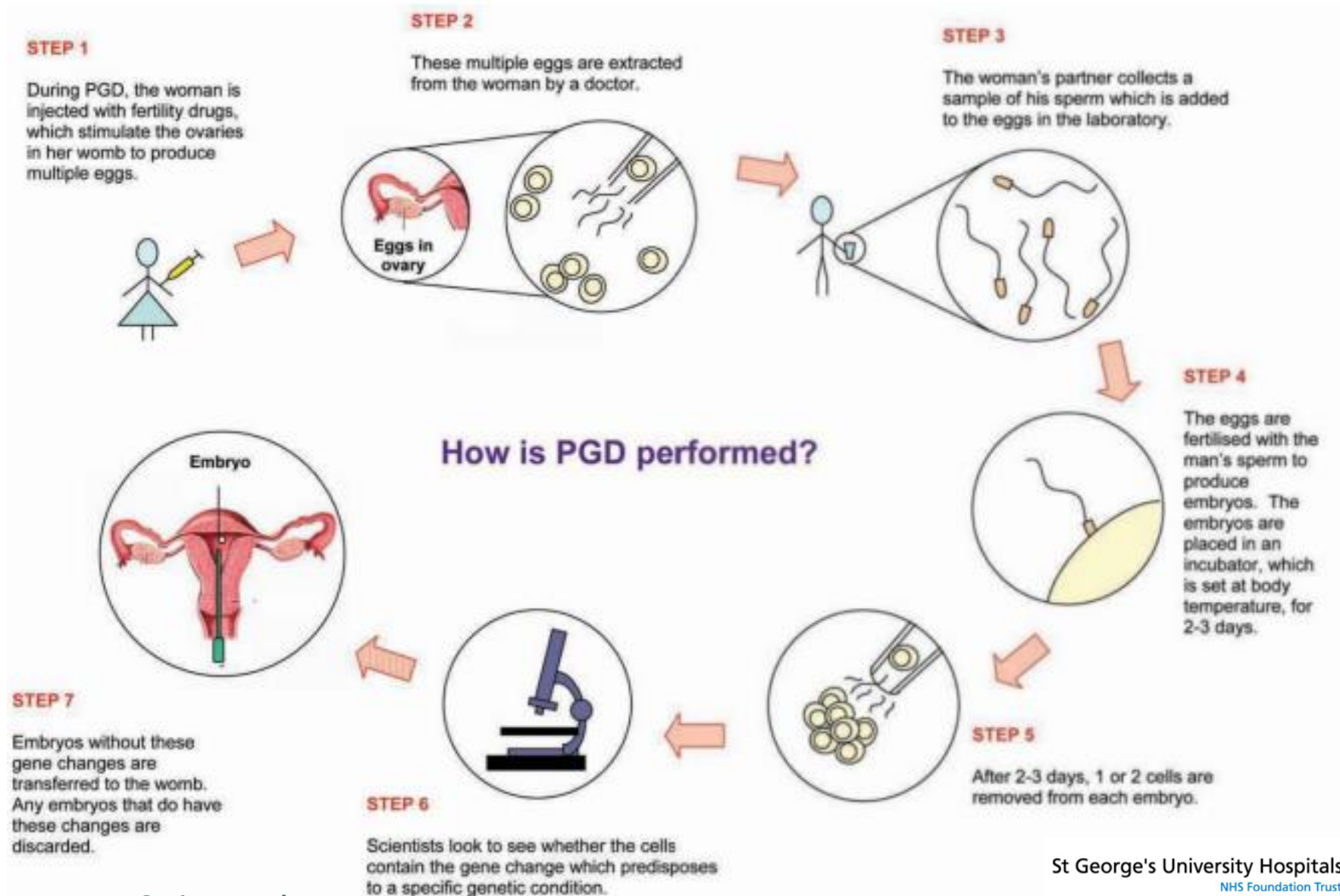


HDA Young adults - PGD

Dr Nayana Lahiri
Consultant in Clinical Genetics
Honorary Senior Lecturer

Preimplantation Genetic Testing (PGT-M)

Appointment
Lab workup
ACU
3-5 months



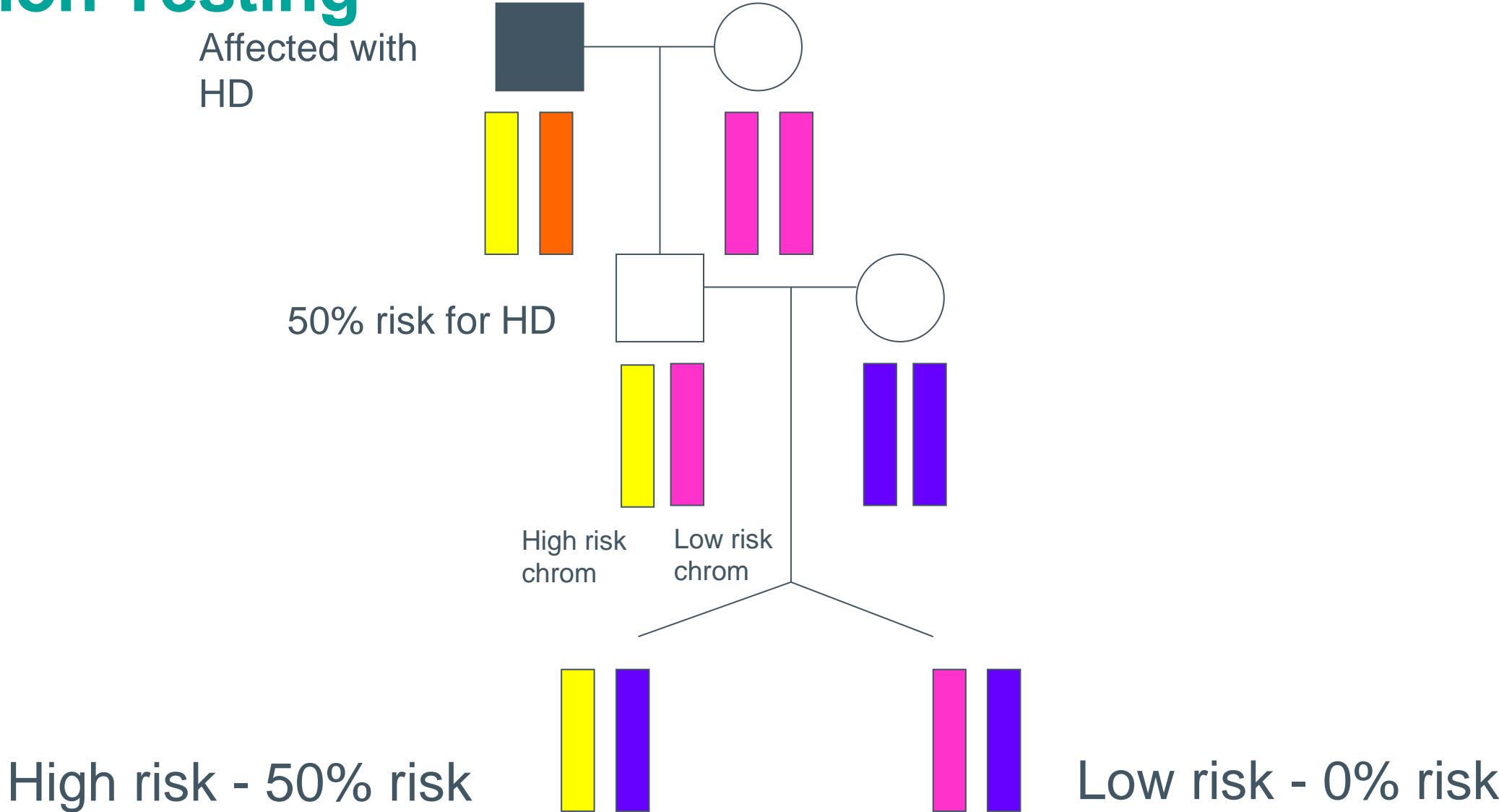
4-9 weeks

2-4 months

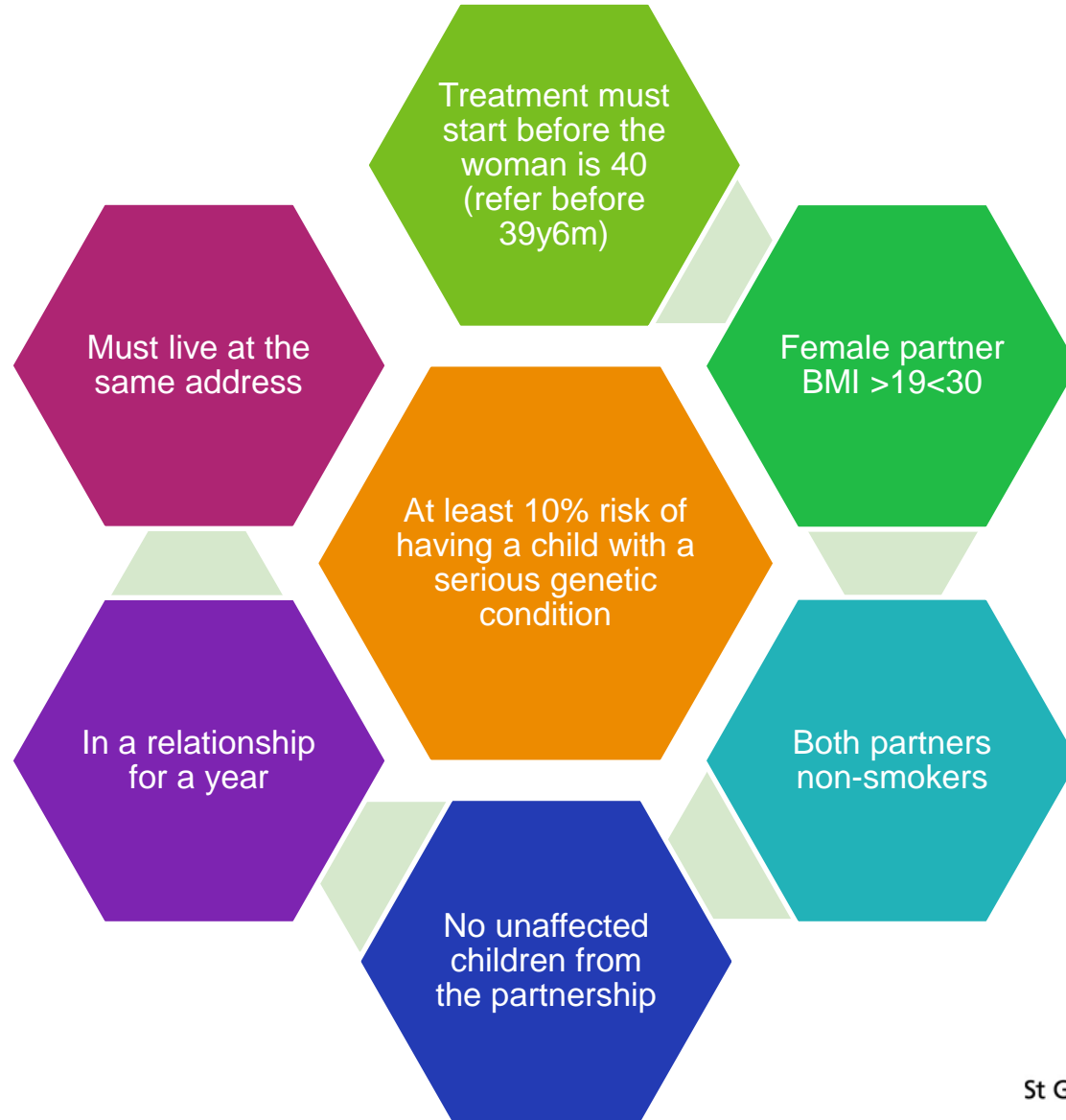
Two ways of testing

- Direct testing
 - Parent has had a positive predictive test
 - Direct test to look for a genetic alteration in an embryo or pregnancy
- Exclusion testing
 - 'At risk' parent has not had a predictive test
 - Sample available from their affected parent

Exclusion Testing



NHS England funding criteria



NHS England funding criteria

- 3 cycles of PGT-M on the NHS
- Funding stops if
 - There is a healthy child
 - Not responding to treatment
 - No longer fit the eligibility criteria
- Private funded cycles – approx. £10-15,000

Things to think about



Where Do I Go?

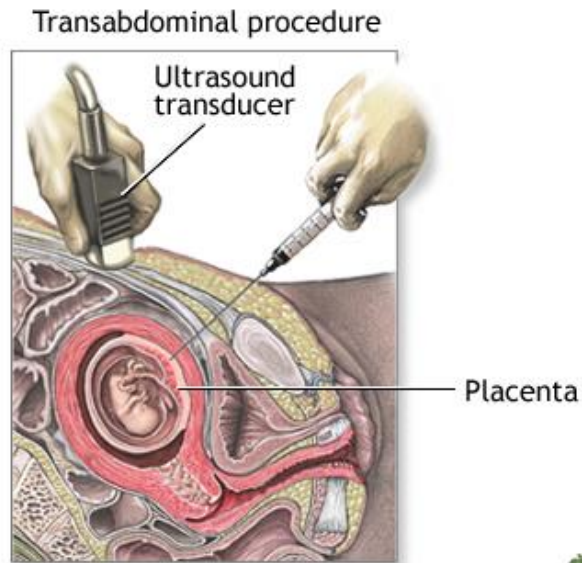
GP referral to your local genetics service – there are 23 of them in the UK

- Guy's
 - Leeds
 - Sheffield
 - Exeter
- CRGH
- Oxford



Prenatal Diagnosis (testing a pregnancy)

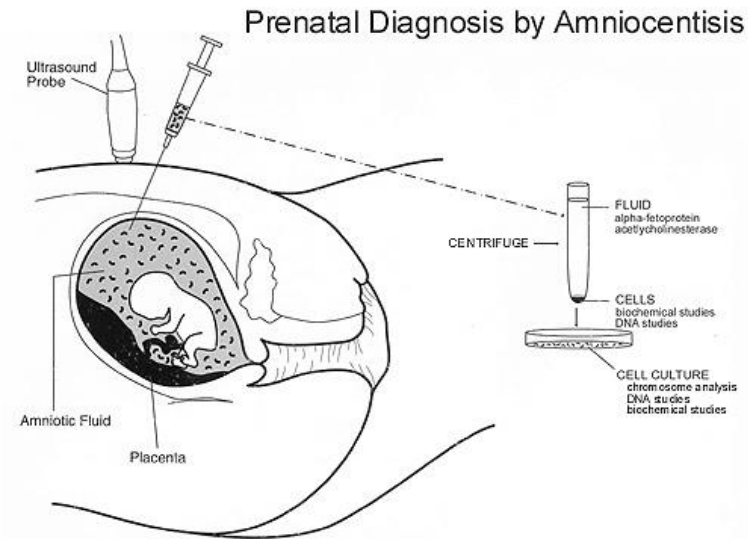
Chorionic Villus Sampling



ADAM.

- 11 weeks of pregnancy
- 1% risk of miscarriage

Amnioscentesis



- 16 weeks of pregnancy
- <1% risk of miscarriage
- Later termination of pregnancy is physically and psychologically more difficult

Resources

- [Huntington's Disease Youth Organization - Having Children \(hdyo.org\)](http://hdyo.org)
- [Pre-implantation genetic testing for monogenic disorders \(PGT-M\) and Pre-implantation genetic testing for chromosomal structural rearrangements \(PGT-SR\) | HFEA](#)
- [Pre-implantation genetic testing \(PGT-M/SR\) - Overview | Guy's and St Thomas' NHS Foundation Trust \(guysandstthomas.nhs.uk\)](http://guysandstthomas.nhs.uk)
- [PGD IVF: Preimplantation Genetic Diagnosis Clinic in London \(crgh.co.uk\)](http://crgh.co.uk)
- [PGT-M Testing | Care Fertility](#)