08.00	Registration, refreshment and exhibition
08.50	Welcome, *Yacoub Khalaf, Consultant in Reproductive Medicine & Surgery & Sub-Specialist in Reproductive Medicine & Surgery, Director of the Assisted Conception Unit & HFEA Person Responsible , Guy's and St Thomas' Hospital*
08.55	Introduction to genetic diseases, their mode of inheritance and their burden on health and reproduction, *Dr Anne Lampe, Consultant in Clinical Genetics, South East Scotland Clinical Genetic Service*
09.15	Reproductive options available to couples at risk, *Janine Elson, Consultant in Gynaecology and Reproductive Medicine, Liverpool Womens Hospital*
09.35	The current status of NIPT and prenatal diagnosis, *Kathy Mann, Lead Clinical Scientist (Prenatal & Reproductive Genetics) Viapath Analytics, Guy's Hospital*
09.55	An introduction to PGT and expanded carrier screening, *Joyce Harper, Professor, University College London*
10.15	PGD counselling and clinical pathway, *Charlotte Tomlinson, Guys & St. Thomas' NHS Trust*
10.35	Refreshment and exhibition
11.00	How does the HFEA regulate PGT?, *Dr Anne Lampe, Consultant in Clinical Genetics, South East Scotland Clinical Genetic Service*
11.30	Embryology of PGT, *Alpesh Doshi, Director of Embryology, Consultant Embryologist, Embryology and PGD Academy*
12.00	PGT diagnostic technologies, *Roy Pascal Naja, Laboratory Director, IGENOMIX UK*
12.30	Lunch and exhibition
13.30	Clinical assessment / preparation of PGT couples and factors that affect success and clinical result, *Tarek El-Toukhy, Consultant Gynaecologist and Sub- specialist in Reproductive Medicine and PGD, Guy's and St Thomas' Hospital NHS Foundation Trust*
14.00	Service delivery models, *Jonathan Skull, Consultant in Reproductive Medicine and Surgery, Clinical Head of Assisted Conception, Jessop Fertility, Sheffield Teaching Hospitals NHS Foundation Trust*
14.20	Case Discussions, *Yacoub Khalaf, Consultant in Reproductive Medicine & Surgery & Sub-Specialist in Reproductive Medicine & Surgery, Director of the Assisted Conception Unit & HFEA Person Responsible , Guy's and St Thomas' Hospital*
15.00	Refreshment and exhibition
15.30	Chair, *Joyce Harper, Professor, University College London* Debate: This house believes that clinical evidence supports routine use of PGT. Against the motion, *Yacoub Khalaf* For the motion, *Roy Pascal Naja*
16.10	Open discussion
16.30	Overall discussion of the day
17.00	Close

PGT, ABSTRACTS AND BIOGRAPHIES

DAY 1

Thursday 06 June

Dr Anne Lampe, Consultant in Clinical Genetics, South East Scotland Clinical Genetic

Dr Anne Lampe is a consultant in Clinical Genetics at the South East of Scotland Clinical Genetic Service in Edinburgh and has a special interest in rare syndrome diagnosis and eye genetics. She provides adult and paediatric genetic services for Fife, including prenatal and predictive testing.
Anne graduated from medical school at the Albert-Ludwigs-University in Freiburg, Germany and completed a PhD researching the role of collagen VI in muscular dystrophy at Newcastle University.
Anne is a Fellow of the Royal College of Physicians (Edinburgh) and is a member of the British Society for Genetic Medicine, the European Society of Human Genetics, the Clinical Genetics Society and the UK Eye Genetics Group. She is also an authority member of the HFEA.

Introduction to genetic diseases, their mode of inheritance and their burden on health and reproduction

Learning points:
Mode of inheritance affects genetic risk
A genetic diagnosis may affect different members of a family unit in different ways

Apart from affecting physical health, genetic disorders can also have a major impact on psychological and social well being of both patients and their families. Using examples from the "Telling Stories Understanding Real Life Genetics" project (http://www.tellingstories.nhs.uk) I will explore how genetic disorders and genetic testing results may affect inter-generational relationships, pose emotional challenges and ethical dilemmas.

Janine Elson, Consultant in Gynaecology and Reproductive Medicine, Liverpool Womens Hospital

Janine is an experienced Consultant in Gynaecology and a Subspecialist in Reproductive Medicine and Surgery. She has run Reproductive Genetics programmes for several large Fertility Groups and teaching hospitals. She was a member of the NHS England PGD policy group, and currently sites on the RCOG Clinical Quality Assurance Group. She was a co author of ESHREs Recurrent Pregnancy Loss guideline.

Reproductive options available to couples at risk

Learning points:
Importance of individualized patient care
Genetic and reproductive counselling is key
Information giving is key

Those at risk of a genetic condition may be aware of this because of screening tests carried out preconception, following pregnancy loss, after the birth of an affected child or because of their individual or family history. The key to managing such patients is counselling regarding the condition at which they are at risk, and a detailed discussion of the options available to them. This presentation will look at who is at risk, and the reproductive options that should be considered in each scenario.

Kathy Mann, Principal Clinical Scientist, Prenatal and Reproductive Genetics, Viapath Analytics, Guy's Hospital

Kathy Mann PhD FRCPath, is lead Clinical Scientist for Prenatal & Reproductive Genetics within the Regional Genetics Laboratory, Guy's Hospital, London. With colleagues she developed the first QF-PCR service in the NHS for the rapid detection of prenatal aneuploidy and is widely published in the field of prenatal QF-PCR analysis including co-

authorship of the UK and European QF-PCR Best Practice Guidelines. She sits on the UK GenQA Specialist Advisory Group for prenatal testing which has published guidelines for the reporting of NIPT results.

The current status of NIPT and prenatal diagnosis

1. NIPT for trisomies is a screening test; confirmation of high risk results by invasive testing is recommended.
2. Evidence supports the use of NIPT for trisomies 13, 18 and 21; the use of NIPT for sex chromosome aneuploidy and microdeletions is not currently recommended and will result in additional non-invasive tests.
3. NIPD results for single gene disorders and fetal sexing are diagnostic and do not require confirmation. NIPD for a growing number of disorders is available from 10 weeks gestation.

Prenatal testing for genetic conditions has undergone a transformation in the UK in recent years; QF-PCR and array CGH technologies have largely replaced FISH and karyotype analysis for the detection of chromosome abnormalities and the long awaited goals of non-invasive prenatal testing and diagnosis (NIPT and NIPD) are finally being realised with an immediate impact on prenatal testing strategies. Whole exome sequencing (WES) is beginning to be applied to prenatal diagnosis with the latest data demonstrating significant clinical utility for a subset of pregnancies. Whilst there is no doubt that these exciting developments have the potential to improve prenatal testing, understanding of the limitations of these tests is necessary to minimise unhelpful and unexpected results. These technologies and their prenatal application will be reviewed with particular focus on current recommended practice.

Charlotte Tomlinson, Guys & St. Thomas' NHS Trust

Charlotte undertook a trainee genetic counsellor position at Bristol clinical genetics service from 2007 to 2009. She then worked as a genetic counsellor at St. George's Hospital clinical genetics service from 2009 to 2016, when she moved to Guy's Hospital clinical genetics service. In September 2018, she moved into the consultant genetic counsellor in general genetics and PGD role. In 2010 she was certified by the Genetic Counsellor Registration Board (GCRB) and re-registered in 2015. She has been a GCRB assessor of portfolios for a number of years and was elected to the GCRB board in 2019. Charlotte is a member of the British Society for Genetic Medicine Fetal Medicine Genomics steering committee.

PGD counselling and clinical pathway

Learning points:
- Learning about the counselling challenges of PGD.
- Understanding the importance of a PGD pathway.
- Close collaboration of the MDT team within the PGD service is essential to addressing the challenges.

The field of PGD has progressed significantly in terms of process and technology over the last 20 years. Whilst we are now able to offer PGD for more genetic conditions, the complexity of these conditions brings their own challenges. This talk will highlight these challenges and how a pathway and close MDT working is essential to managing these complexities.

Anne Lampe, Consultant in Clinical Genetics, South East Scotland Clinical Genetic Service

Biography as above

How does the HFEA regulate PGT?

What the law says about PGD and PGS
The regulatory pathway for PGD from patient consultation to embryo transfer
The HFEA's position on PGS

PGD is regulated by the Human Fertilisation and Embryology Act 1990 (as amended)
A clinic must have a licence from the HFEA to carry out PGD testing. Then PGD can be carried out for a heritable condition for specific purposes listed in the Act
- where there is a particular risk that the embryo to be tested may have a genetic, chromosomal or mitochondrial abnormality,
- where there is a particular risk that any resulting child will have or develop a gender related serious disability, illness or medical condition.
The regulatory pathway from patient consultation to embryo transfer will be discussed.

Roy Pascal Naja, Laboratory Director, IGENOMIX UK

Dr Naja has a PhD in Human Genetics from McGill University, Montreal, Canada. Before joining Igenomix UK as Laboratory Director, Dr Naja was a Principal Clinical Scientist at the Neurogenetics laboratory UCLH/NHS foundation trust in London. From Dec 2015 to Aug 2016, Dr Naja was the senior Clinical Scientist at Reprogenetics UK. From Oct 2013 to Dec 2015 Dr. Naja was the Laboratory Manager of the UCL Centre for PGD. Dr Naja is a Clinical Scientist (HCPC) and an"Diplomate" member of the Royal College of Pathologists. Dr Naja is registered with UKAS as a technical assessor.

PGT diagnostic technologies

Learning points:

1. PGT diagnostic technologies have evolved over the last three decades and continue to evolve.
2. High resolution NGS has revealed chromosomal mosaicism in TE biopsies.
3. Rigorous validation of PGT procedures is mandatory.

PGT diagnostic technologies. Abstract: Preimplantation Genetic Testing (PGT) includes testing for monogenic disorders (PGT-M), structural rearrangements (PGT-SR) and aneuploidies (PGT-A). Technologies used in PGT-A/SR have evolved over the years from covering a single or few chromosomes by using Fluorescent In Situ Hybridization (FISH) to high resolution 24-chromosome screening by using Next Generation Sequencing (NGS). The increased sensitivity of NGS has revealed chromosomal mosaicism in TE biopsies thus creating some uncertainty regarding embryo selection for transfer. Similarly, methods used for PGT-M have evolved from interrogating a single mutation by using direct PCR to detecting the inheritance of any gene by using genome-wide linkage information derived from SNP arrays (Karyomapping method, Illumina) or NGS (Haplarhythmisis/onePGT, Agilent). All preimplantation tests must be validated by the diagnostic laboratory before use to ensure high analytical accuracy as they generally involve small amounts of sample DNA as starting material.

Tarek El-Toukhy, Consultant Gynaecologist and Sub- specialist in Reproductive Medicine and PGD, Guy's and St Thomas' Hospital NHS Foundation Trust

Tarek El-Toukhy qualified in 1991. He completed a Masters degree and an MD degree in Gynaecology. He completed the RCOG accredited subspecialty training in Reproductive Medicine and Surgery at Guy's and St. Thomas' Hospital NHS Foundation Trust, where he was appointed as a consultant in Reproductive Medicine and Surgery and Pre-implantation Genetic Diagnosis (PGD). His special interests are PGD, recurrent implantation failure, hysteroscopic surgery and prevention of OHSS. He is a scientific editor for the British Journal of Obstetrics and Gynaecology

DAY 1

Thursday 06 June

Clinical assessment/ preparation of PGT couples and factors that affect success and clinical results

Key Learning Points:

1. Understand the clinical assessment needed for PGD couples
2. Review the preparatory steps required before starting a PGD cycle
3. Explore the factors influencing the outcome of PGD cycle

Pre-implantation Genetic Diagnosis (PGD) was developed in the late 1980s as an alternative to prenatal diagnosis for couples at substantial risk of conceiving a pregnancy affected by a known genetic disorder. It enables IVF clinics to select embryos for implantation so that at-risk families can avoid passing on genetic disease to their children and to subsequent generations. Over the past decade the use of PGD has increased as its indications have expanded and changed, both with demand and improvement in molecular diagnostic techniques. The talk will cover clinical assessment and preparatory steps required before starting a PGD cycle, emphasize the difference between PGD and pre-implantation genetic screening (PGS), and explore the factors influencing the outcome of PGD cycle. Clinical results will also be presented.

Jonathan Skull, Consultant in Reproductive Medicine and Surgery, Clinical Head of Assisted Conception, Jessop Fertility, Sheffield Teaching Hospitals NHS Foundation Trust

Jonathan qualified from Bristol University and undertook postgraduate training in Sheffield and London. He was the Senior IVF Co-ordinator at the Hammersmith before returning to Sheffield in

1998, first as a Lecturer and then as Consultant. He was instrumental in establishing the Assisted Conception Unit at the Jessop Wing, where he has been the Clinical Head of the unit since it opened in 2001. He is also the lead clinician for the PGD satellite service. His other interests include Minimal Access surgery as well as active involvement with postgraduate training in Reproductive Medicine.

Service delivery models

Learning points:
1. Satellite PGD services allow patients to receive the majority of their treatment closer to home making the whole procedure more convenient and less stressful.
2. Various models for running a satellite PGD service will be discussed.
3. Good links with local genetic services and communication with the main PGD centre are essential for a successful programme Importance of good communication.

Service Delivery Models. Jessop Fertility started working with Guy's as a Satellite PGD centre in 2008 and the demand for the service has increased significantly. Patients are initially referred to the service after initial assessment by the local Genetics team, where their suitability for PGD is assessed and relevant samples sent for work up. A full fertility assessment is undertaken concurrently in the fertility clinic to ensure that IVF+PGD is appropriate for the couple. Patients are then seen in a Joint PGD clinic to plan their treatment. All IVF monitoring is conducted locally with patients only having to travel to the PGD centre for the egg collection and subsequent frozen transfer. Alternative satellite PGD models where patients could undertake a local embryo biopsy with genetic testing performed at another centre will also be discussed. For satellite programmes to be successful there has to be excellent communication between all relevant disciplines.