Welcome to this is my
Health Screening and Ultrasound Centres
A New Combined Screening Test

‘NIPT & Ultrasound’

Judith Pilling, DCR(R) DMU MHSc,
Managing Director, this is my: Ltd
Professor Howard Cuckle D Phil, MSc BA
Adjunct Professor of Obstetrics and Gynaecology, Columbia University, New York, USA. He is also Emeritus Professor at the School of Medicine at the University of Leeds and a director of this is my Ltd.

Indera Sehmi
Lead Screening Coordinator and Corporate Manager

Mr Gerald Mason
MBChB, FRCOG, MRCOG, MD
High Risk obstetrician in Feto Maternal Medicine retired from Leeds General Infirmary. Also, and a director of this is my Ltd.

Judith Pilling DCR(R) DMU MHSc
Director of this is my: Health Screening & Ultrasound Centres and Expert Clinical Specialist.

2003 Leeds Screening Centre

Leeds Antenatal Screening Service

Leeds Ultrasound Screening Service

Biochemistry

Ultrasound
Team of 30 covering 8 screening locations offering full range of pregnancy services including NIPT

Leeds (inc Head Office)
Manchester
Hull
Halifax
London : Harley Street
London Canary Wharf
Durham
Liverpool
Brief Screening History

- **1991**: 2\(^{nd}\) Trimester: Triple (NHS) / Triple+ (AFP / UE3 / HCg)
- **1997**: 2\(^{nd}\) Trimester: Biomark / Quad: (/ AFP / UE3 / HCg / InhibinA)
- **1997**: Fragments of fetal DNA found in maternal blood (Dennis Lo)!
- **1998**: Primark: 1\(^{st}\) Trimester: (NT / PAPP A / AFP / HCg / UE3)
- **2003**: Leeds Screening Centre Opened: LASS & LUSS
- **2005**: Addmark 1\(^{st}\) Trimester: (NT / NB / PAPP A / AFP / HCg / UE3 + AFP / UE3 / HCg / InhibinA)
- **2005**: Genmark: 2\(^{nd}\) Trimester: (NT or NF / NBL / AFP / UE3 / HCg / InhibinA)
- **~2008**: Cell Free DNA using ‘Next Generation Sequencing’
- **2010**: Leeds Screening Centre became **this is my:ltd**
- **2011**: First Test Available in USA
- **2013**: this is my offered Non-Invasive Prenatal Testing (NIPT) 10+ weeks
Screening Options

1\textsuperscript{st} Trimester (11 – 13 weeks)
- Addmark ®
- Nuchal +
- NHS combined

2\textsuperscript{nd} Trimester (14 + weeks)
- Genmark ™
- NHS Quadruple test

Now!!
Non-Invasive Prenatal Testing (NIPT) 10+ weeks
## Down’s syndrome screening

<table>
<thead>
<tr>
<th>TEST</th>
<th>Gestation</th>
<th>DR %</th>
<th>PR %</th>
</tr>
</thead>
<tbody>
<tr>
<td>ADDMARK</td>
<td>9 – 13</td>
<td>93</td>
<td>0.6</td>
</tr>
<tr>
<td>NUCHAL +</td>
<td>11 – 13</td>
<td>89</td>
<td>2.8</td>
</tr>
<tr>
<td>GENMARK</td>
<td>14 – 20</td>
<td>80</td>
<td>2.7</td>
</tr>
<tr>
<td>NHS COMBINED</td>
<td>11 – 13</td>
<td>75 - 90</td>
<td>2 - 3</td>
</tr>
<tr>
<td>NHS QUAD</td>
<td>14 – 20</td>
<td>75</td>
<td>3</td>
</tr>
<tr>
<td>NIPT</td>
<td>10+</td>
<td>99</td>
<td>&lt;0.2</td>
</tr>
</tbody>
</table>
Non Invasive Prenatal Testing

<table>
<thead>
<tr>
<th>Company</th>
<th>Test Name</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ariosa / Roche / TDL</td>
<td>Harmony</td>
</tr>
<tr>
<td>BGI Healthcare</td>
<td>NIFTY</td>
</tr>
<tr>
<td>Genesis Cooper</td>
<td>Serenity</td>
</tr>
<tr>
<td>Natera</td>
<td>Panorama</td>
</tr>
<tr>
<td>Premaithia</td>
<td>Iona</td>
</tr>
</tbody>
</table>

T21 >99% accurate
Ultrasound Imaging & Non Invasive Prenatal Testing
Ultrasound Imaging

6 weeks

10 weeks
Ultrasound Imaging

Viability
Fetal Number & Chronicity
Gestational Age
Fetal Anatomy
1st Trimester Fetal Anatomy
  Head
  Heart
  Arms
  Stomach
  Cord Insertion
  Bladder
  Legs

1st Trimester Chromosomal ‘Markers’
  Nuchal Translucency
  Nasal Bone
  Ductus Venosus
  Tricuspid Valve Regurgitation
  Facial Angle
  Intra Cranial Translucency
Nuchal Translucency

Normal

Increased
Down’s syndrome screening
Nasal Bone

Present

Absent
Down’s syndrome screening
Ductus Venosus

Normal

Abnormal
Down’s syndrome screening
Tricuspid Valve

Normal

Abnormal
Down's syndrome screening
Facial Angle

Normal

Abnormal
Intracranial translucency
Risk of Spina bifida
Non Invasive Prenatal Testing (NIPT)

NIPT is a prenatal screening test that can be performed beginning around the 10th week of pregnancy.

Noninvasive Prenatal Testing

Fetus
Placenta

Small fragments of cell-free DNA from the placenta enter the mother's bloodstream.

Mother's Bloodstream

Placental (“fetal”) DNA
Maternal DNA

Cell-free DNA in a sample of the mother’s blood is analyzed for evidence of extra or missing fetal DNA segments.
NIPT: Simple, Fast, Reliable, SAFE.

How Does Serenity NIPT Work?

Results: 3 – 10 working days (average: 5)
Depending on processing laboratory
NIPT Report: Low Risk

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**Prenatal Aneuploidy Test Report**

**Provider Information**
- **Name:** Mr. Gerald Mason
- **Address:** This is my LS1115104 93 Water Lane Leeds LS115QN United Kingdom
- **Phone:** 08452729999/01132621675
- **Email:** info@thisismy.co.uk judith.pilling@thisismy.co.uk

**Test/Platform:** NGS sequencing
**Primary sample type:** Cell free DNA from maternal plasma

**Prenatal aneuploidy test results - Singleton pregnancy**

<table>
<thead>
<tr>
<th>CHROMOSOMES</th>
<th>RESULTS</th>
<th>INTERPRETATION</th>
</tr>
</thead>
<tbody>
<tr>
<td>Single Olsen 13</td>
<td>No aneuploidy detected</td>
<td>Results consistent with two copies of chromosome 13</td>
</tr>
<tr>
<td>Single Olsen 18</td>
<td>No aneuploidy detected</td>
<td>Results consistent with two copies of chromosome 18</td>
</tr>
<tr>
<td>Single Olsen 21</td>
<td>No aneuploidy detected</td>
<td>Results consistent with two copies of chromosome 21</td>
</tr>
<tr>
<td>Sex chromosomes</td>
<td>No aneuploidy detected</td>
<td>Results consistent with two copies of sex chromosomes XX</td>
</tr>
</tbody>
</table>

**Comments:** This is a screening test; therefore, false positive and false negative results can occur. Clinical correlation is indicated. If definitive diagnosis is desired, chorionic villus sampling or amniocentesis would be necessary.

**Fetal karyogram:** 11%

**Sample ID:**
- **Sample ID:** TM-02072
- **Draw date:** 05 Jun 2017
- **Receipt date:** 06 Jun 2017
- **Report time and date:** 09 Jun 2017 14:30:00

**Limitations of Test:**
- This test is designed to screen for chromosome aneuploidies and is validated for chromosomes 13, 18, 21, X and Y. The test is validised for singleton and twin pregnancies with gestational age of at least 10 weeks 0 days, as estimated by last menstrual period, crown rump length, or other appropriate method (normal to eight weeks’ gestation as determined by ultrasound). These results do not eliminate the possibility that this pregnancy may be associated with other chromosomal or subchromosomal abnormalities, birth defects, and other conditions.
- When an aneuploidy detected result is reported in a twin pregnancy, the status of each individual fetus cannot be determined. Although the presence or absence of Y chromosome material can be reported in a twin pregnancy, the occurrence of sex chromosome aneuploidies such as X0, XXX, XXXY, and XXY cannot be evaluated in twin pregnancies.
- There is a small possibility that the test results might not reflect the chromosomes of the fetus, but may reflect chromosomal changes of the placenta (maternal placental mosaicism) or of the mother (chromosomal mosaicism). Follow up diagnostic testing is recommended for any positive result finding.

**Performance Data Table**

<table>
<thead>
<tr>
<th>Anomaly</th>
<th>Sensitivity</th>
<th>Specificity</th>
</tr>
</thead>
<tbody>
<tr>
<td>Chromosome 13</td>
<td>95.0%</td>
<td>99.4%</td>
</tr>
<tr>
<td>Chromosome 18</td>
<td>98.3%</td>
<td>99.50%</td>
</tr>
<tr>
<td>Chromosome 21</td>
<td>93.4%</td>
<td>99.50%</td>
</tr>
<tr>
<td>XXY</td>
<td>97.0%</td>
<td>99.0%</td>
</tr>
<tr>
<td>XY</td>
<td>99.0%</td>
<td>99.0%</td>
</tr>
</tbody>
</table>

**Disclaimer:**
- The manner in which this information is used to guide patient care is the responsibility of the healthcare provider, including advising for the need for genetic counseling or diagnostic testing. Any test should be interpreted in the context of all available clinical findings. The test was developed by Cooper Genetics UK Ltd, which is powered and licensed by Illumina with NextSure technology. In exceptional circumstances, such as high demand, samples may occasionally be processed at Illumina Health Inc., wholly owned by Illumina Inc. California, USA. CAP 758532 CLIA 5020053591

**Lab Director:** Tony Gordon

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Croydon, Surrey, CR9 1BY
Tel: +44 (0) 20 7891 2084

**Accredited to ISO 15189:2012**
NIPT Report: High Risk
Reason why clients opt for NIPT

- increased risk from prior screening (25%)
- advanced maternal age
- maternal anxiety
- previous affected pregnancy
- failed NT
NIPT Screens Performed n= 5500
July 2013 – June 2017

Maternal age range: 19-53 years (m=36 years)

Gestational age range: 10-38 weeks (m=13+4)

65% of samples were taken at 14 weeks or less

90% of clients request gender information