

ANTENATAL CLINIC NHS Trust
BURNLEY GENERAL HOSPITAL
CASTERTON AVENUE
BURNLEY
LANCASHIRE BB10 2P0

Telephone 01282 804374 Patient Hospital No:

NHS NO

30th April 2019

Dear

We are writing to inform you that the screening test taken on Off shows that you are in the lower chance group for having a babies with Down's Syndrome.

This is a screening test only and not a diagnosis.

The chance of Down's Syndrome is Twin One 1: NUMBER
Twin Two 1: NUMBER

This means that Twin 1 has one chance in Number that the baby has the condition.

This means that Twin 2 has one chance in Number that the baby has the condition.

The chance of twin 1 and twin 2 with combined Edwards' and Patau's syndrome is 1 chance in $NU \cap VOI$

We would not normally offer further testing with these results, however, if you have any further queries please do not hesitate to contact us.

Please show this letter to your midwife at your next visit. Yours sincerely







ANTENATAL CLINIC NHS Trust
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This means that Twin 1 has one chance in Number that the baby has the condition.
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We would not normally offer further testing with these results, however, if you have any further queries please do not hesitate to contact us.

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ANTENATAL CLINIC NHS Trust
BURNLEY GENERAL HOSPITAL
CASTERTON AVENUE
BURNLEY
LANCASHIRE BB10 2PQ

Telephone 01282 804374 Patient Hospital No:

NHS NO.

30th April 2019

Dear

Combined screening for Edwards' and Patau's Syndromes

We are writing to inform you that the screening performed on DATE shows that you are in the lower chance group for having a baby with Edwards' and Patau's Syndromes.

This is a screening test only and not a diagnosis.

The chance of Edwards' and Patau's syndrome is 1: NUMbo/ this means there is one chance in NUMbo/ that the baby has the condition.

We would not normally offer further testing with these results, however, if you have any further queries please do not hesitate to contact us.

Please show this letter to your midwife at your next visit. Yours sincerely







ANTENATAL CLINIC NHS Trust
BURNLEY GENERAL HOSPITAL
CASTERTON AVENUE
BURNLEY
LANCASHIRE BB10 2PQ

Telephone 01282 804374 Patient Hospital No

NHS NO.

30th April 2019

Dear

Combined screening for Down's Syndrome, Edwards' and Patau's Syndromes

We are writing to inform you that the screening performed on DATE shows that you are in the lower chance group for having a baby with Down's Syndrome, Edwards' and Patau's syndromes.

This is a screening test only and not a diagnosis.

The chance of Down's Syndrome is 1:Number this means that there is one chance in number that the baby has the condition.

The chance of Edwards' and Patau's syndromes is $1:\Lambda \lor mb \lor f$ which means that there is one chance in $\Lambda \lor f b \lor f$ that the baby has the condtion.

We would not normally offer further testing with these results, however, if you have any further queries please do not hesitate to contact us.

Please show this letter to your midwife at your next visit.

Yours sincerely







ANTENATAL CLINIC NHS Trust
BURNLEY GENERAL HOSPITAL
CASTERTON AVENUE
BURNLEY
LANCASHIRE BB10 2PQ

Telephone 01282 804374 Patient Hospital No:

NHS NO

30th April 2019

Dear

Maternal Serum Screening for Down's Syndrome

We are writing to inform you that the quadruple blood test sample taken on DATE shows that you are in the lower chance group for having a baby with Down's Syndrome.

This is a screening test only and not a diagnosis.

The chance of Down's Syndrome is 1: n where this means that there is one chance in n whose that the baby has the condition.

We would not normally offer further testing with these results, however, if you have any further queries please do not hesitate to contact us.

Please show this letter to your midwife at your next visit. Yours sincerely







ANTENATAL CLINIC NHS Trust
BURNLEY GENERAL HOSPITAL
CASTERTON AVENUE
BURNLEY
LANCASHIRE BB10 2P0

Telephone 01282 804374 Patient Hospital No:

NHS NO

30th April 2019

Dear

Combined screening for Down's Syndrome

We are writing to inform you that the screening performed on DATE shows that you are in the lower chance group for having a baby with Down's Syndrome.

This is a screening test only and not a diagnosis.

The chance of Down's Syndrome is 1: NVMDOF this means that there is one chance in NVMDOF that the baby has the condition.

We would not normally offer further testing with these results, however, if you have any further queries please do not hesitate to contact us.

Please show this letter to your midwife at your next visit.

Yours sincerely









Lancashire Women & Newborn Centre Casterton Avenue Burnley

BB10 2PQ

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Screening co-ordinator: Janyce Sutherland Tel: 01282 803019 Email: Janyce.Sutherland@elht.nhs.uk

> Hospital N NHS No.: Date 09/0

Non-Invasive Prenatal Testing (NIPT) Results

Patient:

Exam date:

Indication

Raised risk on combined screening

History

Risk 1:110 of trisomy 21

Pregnancy

Singleton pregnancy. Number of fetuses: 1.

Test Results

Please find enclosed the results of your recent Harmony Non-Invasive Prenatal Test (NIPT).

The results indicate that you are LOW PROBABILITY for Trisomy 21 (Down Syndrome), Trisomy 18 (Edward Syndrome) and Trisomy 13 (Patau Syndrome).

The screen for sex chromosome problems has come back as LOW PROBABILITY.

The fetal sex was determined to be MALE.

The amount of fetal cells within the blood test was deemed sufficient to analyse the sample (fetal fraction 21%).

As you recall, we discussed that the NIPT is not a diagnostic test so will not detect all cases of chromosome problems. Only invasive tests (amniocentesis) are diagnostic at this stage. If you wish to proceed with an invasive test at this stage please contact the screening team.

The test will not detect mosaicism, partial chromosome problems, translocations or maternal chromosomal problems. Harmony does not detect structural problems with your baby - these are best detected at your 20-week anomaly scan.

Follow-up

No further screening tests have been arranged at this stage.

If you wish to proceed with an invasive test (amniocentesis) please contact the screening midwives on the number above and they will arrange this for you in the Fetal Medicine Unit.

If you wish to discuss the test any further please feel free to contact my secretary on 01282-803197.

Consultant Obstetrician







Lancashire Women & Newborn Centre Casterton Avenue Burnley BB10 2PQ

Screening co-ordinator: Janyce Sutherland Tel: 01282 803019 Email: Janyce.Sutherland@elht.nhs.uk

> Hospital No NHS No. Date 0!

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Non-invasive Prenatal Testing (NIPT)

Patient:

Exam date:

Indication

Raised risk on combined screening

History

Risk 1:110 of trisomy 21

Pregnancy

Singleton pregnancy. Number of fetuses: 1.

Counselling Summary

Attended for Non-Invasive Prenatal Testing (NIPT) due to raised risk of trisomy 21 on

combined screening.

Explained that this is a private test not available on NHS at present.

Harmony is the company of choice in ELHT, but other providers are available privately. Cost

of the test is £350 (Harmony will invoice you for this directly).

Discussed that screening for fetal chromosomal disorders is optional. Implications for the test results discussed. The decision to proceed is based on how each individual perceives the benefits of obtaining the information about chromosomal problems when weighed against the potential emotional and physical risks of testing.

NIPT is a screening test. It cannot diagnose or eliminate the chance that a fetus has a particular chromosomal condition; rather it separates women into high and low risk categories. Testing is performed using a blood test of mother from the arm and is not associated with an increased risk of miscarriage.

We discussed that the only diagnostic test at this gestation is an amniocentesis test. This is a test where a small needle is placed through your abdomen under ultrasound guidance and a small amount of fluid surrounding the baby is taken for analysis. This will confirm if the baby is affected by a chromosomal condition. There is a small risk of 0.5-1% of

miscarriage.

Clinical features of each condition screened for in the test discussed. These included in the

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- Trisomy 21 (Down Syndrome)
- Trisomy 18 (Edwards Syndrome)
- Trisomy 13 (Patau Syndrome)
- Sex chromosome aneuploidies (monosomy X, XXY, XXX, XYY) . The test for this is optional You decided that you want to proceed with this test.
- Sex of the fetus. This test is optional. You have expressed that you wish this test to be performed.

Specific testing methods discussed – there are fragments of chromosomes that circulate outside of cells within the mothers blood. These DNA fragments are referred to as 'cell-free'. Most cell free DNA fragments derive from the mothers blood cells, but some come from the placenta. Because the fetus and placenta derive from the same fertilised cell, they are usually genetically identical. The sequences of cell free DNA fragments are analysed to establish how many fragments there are and which chromosomes they have come from. The laboratory counts the number of fragments from each chromosome of interest, and determines if there are more or less than expected. If, for example, there are more than expected number of fragments from chromosome 21, this indicates an increased chance of the baby having trisomy 21.

The results will be communicated from the test laboratory to East Lancashire Hospitals and communicated to you once this has been received. A result is normally available in 3-5 working days, but can sometimes be longer than this. If you do not hear anything back after 7 working days please contact Mr Maher's secretary or the screening team.

in 3 out of 100 women a repeat test will be required (a result is achieved in approximately two thirds of these). This is because the amount of fetal blood cells in your circulation is not enough for the laboratory to test. This is called the fetal fraction. If the test requires repeating you will not be charged for this.

NIPT is not diagnostic but a screening test. This means that the test has a certain sensitivity for each condition and can sometimes be wrong.

We discussed the sensitivity for Trisomy 21 is over 99% (false positive rate <0.1%)

Trisomy 18 is 97.4% (false positive rate <0.1%)

Trisomy 13 is 93.8% (false positive rate <0.1%)

I explained that the result will give a high or low risk result. If you wish to consider invasive/diagnostic tests following the results these can be arranged in the Fetal Medicine Unit.

Following counselling today, you wished to proceed with the test and written consent was obtained for this.

Ultrasound scan confirms a viable pregnancy.

Blood samples have been taken today using an aseptic non-touch technique and sent by recorded mail to the Harmony laboratory.

Follow-up

Results will normally be available in 3-5 working days and will be communicated to you yie the screening team/Mr Maher.

Consultant Obstetrician

Page 2 of 3 for report of patient







TOI GENETICS

performed to the UK

The Halo Building 1 Mabledon Place London WC1H 9AX

T. +44 (0) 20 7207 7409 E: war-magisterscopica

www.tdlpathalogy.com

Padan and Phoyotal Information PATIENT HAME: DATE OF BIRTING (00/884/0911) MPH

LAPÓRATORY ID OTHER IO

GESTATIONAL AGE:

13 wks 3 days POFFETUSES: IVE STATUSE

non-IVF pregnancy COLLECTION DATE (DD/MM/YYYY) RECEIVED DATE (OD/NIM/YYYY): 29/03/2019 01/04/2019

ACCOUNT V EASTLANC CLINIC NAME! East Lancashire Hospitals Trust REFERRING/ONDERING CUNICIAN: referbing/ordering clinician email DTHER CUBICIAN. OTHER CLINICIAN CONTACT INFORMATION:

REPORT DATE: 04/04/2019

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CHROMOSOME	RESULT	PROBABILITY	RECOMMENDATION
Trisomy 21 (T21)	Low Probability	less than 1/10,000 (0,01%)	Review results with patient
Trisamy 18 (T18)	Low Probability	Less than 1/10,000 (0.01%)	Review results with patient
Trisomy 13 (T13)	Low Probability	Less than 1/10,000 (0.01%)	Review results with patient

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TEST DESCRIPTION

TEST DESCRIPTION

The Narmony Prenatal Test® measures the relative proportion of chromosomes to aid in the assessment of letal instomes 21, 18, and 13. Narmony® parforms a directed analysis of cell-free DNA (cfDNA) in maternal blood and incorporates the fetal fraction of cfDNA in lest results. Test results also incorporate maternal age (or egg donor age) and gestational age related probability based on information previded on the test requisition form. Probability of Jess than 1% is defined as low probability and 1% or greater is defined as high probability. Harmony has been validated in singleton and twin preparactes of at least 10 weeks gestational age. Harmony is not wildstated for use in preparactes with more than two fetuses, demised twin, mosalcisin, partial thromosome aneuploidy, transfortions, maternal aneuploidy, transfortions, maternal aneuploidy, transfortions, maternal aneuploidy, transfortions, maternal aneuploid, transfortions of detect neural tuba defects. Twin results reliect the probability that the pregnancy involves all less one affected fetus. Analysis of cfDNA does not always correlate with fetal genotype, not all aneuploid fetuses will have a high probability result and some audoid fetuses will have a high probability result.

Fetal Sex test quantifies the Y chromosome. A "lemale" result indicates no detection of Y chromosome and a "male" result indicates detection of Y chromosome. It does not exclude sex chromosome anauploidy. For twin pregnancies, a male result indicates one or two mole fetuses.

pregnancies, a mala result indicates one or two mole fetuses.

Sex Chromosome Aneupladdy (SCA) Panel measures proportions of the X and Y chromosomes. Sex chromosome conditions (Monosomy X, XXY, XXY, XXX, XXY) are reported at propoblilies of 1% or greater. An 23Y or XXY result indicates two or more fetal Y chromosomes. Sex Chromosome Aneusloidy Panel has only been validated in singleton pregnancies.

cunical validation data from the harmony productinisert.

	Daletion Hale	Falce Positive Hate
T21	167/[58	0/641
T18	29/19	0/641
тіз	12/12	0/541

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Fetal Sex:

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NHS Trust Lancashire Women & Newborn Centre Casterton Avenue Burnley 8810 2PQ

> Hospital No NHS No.: Date 16/04/2019

Genetic Counselling

Patient:

Exam date:

Indication

Maternal request

Pregnancy

Singleton pregnancy. Number of fetuses: 1.

Test Results

Please find enclosed the results of your recent Harmony Non-Invasive Prenatal Test (NIPT)

The results indicate that you are LOW PROBABILITY for Trisomy 21 (Down Syndrome), Trisomy 18 (Edward Syndrome) and Trisomy 13 (Patau Syndrome).

The fetal sex was determined to be MALE.

The amount of fetal cells within the blood test was deemed sufficient to analyse the sample (fetal fraction 9.7%).

As you recall, we discussed that the NIPT is not a diagnostic test so will not detect all cases of chromosome problems. Only invasive tests (amniocentesis) are diagnostic at this stage. If you wish to proceed with an invasive test at this stage please contact the screening team.

The test will not detect mosaicism, partial chromosome problems, translocations or maternal chromosomal problems. Harmony does not detect structural problems with your baby - these are best detected at your 20-week anomaly scan.

Follow-up

No further screening tests have been arranged at this stage.

if you wish to proceed with an invasive test (amniocentesis) please contact the screening midwives on the number above and they will arrange this for you in the Fetal Medicine

Unit.

If you wish to discuss the test any further please feel free to contact my secretary on 01282-803197.

.Consultant Obstetrician

CC:



NHS Trust
Lancashire Women & Newborn Centre
Casterton Avenue
Burnley
BB10 2PQ

Hospital No: NHS No.: Date 10/6

Genetic Counselling

Patient:

Exam date:

Indication

Maternal request

Pregnancy

Singleton pregnancy. Number of fetuses: 1.

Counselling Summary Attended for Non-Invasive Prenatal Testing (NIPT) due to maternal request

Explained that this is a private test not available on NHS at present.

Harmony is the company of choice in ELHT, but other providers are available privately. Cost of the test is £350 (Harmony will invoice you for this directly).

Discussed that screening for fetal chromosomal disorders is optional. Implications for the test results discussed. The decision to proceed is based on how each individual perceives the benefits of obtaining the information about chromosomal problems when weighed against the potential emotional and physical risks of testing.

NIPT is a screening test. It cannot diagnose or eliminate the chance that a fetus has a particular chromosomal condition; rather it separates women into high and low risk categories. Testing is performed using a blood test of mother from the arm and is not associated with an increased risk of miscarriage.

We discussed that the only diagnostic test at this gestation is an amniocentesis test. This is a test where a small needle is placed through your abdomen under ultrasound guidance and a small amount of fluid surrounding the baby is taken for analysis. This will confirm if the baby is affected by a chromosomal condition. There is a small risk of 0.5-1% of miscarriage.

· Clinical features of each condition screened for in the test discussed. These included:

- Trisomy 21 (Down Syndrome)
- Trisomy 18 (Edwards Syndrome)
- Trisomy 13 (Patau Syndrome)
- Sex chromosome aneuploidies (monosomy X, XXY, XXX, XYY) . The test for this is optional. You decided that you did not want to proceed with this test.
- Sex of the fetus. This test is optional. You have expressed that you wish this test to be performed.

Specific testing methods discussed — there are fragments of chromosomes that circulate outside of cells within the mothers blood. These DNA fragments are referred to as 'cell-free'. Most cell free DNA fragments derive from the mothers blood cells, but some come from the placenta. Because the fetus and placenta derive from the same fertilised cell, they are usually genetically identical. The sequences of cell free DNA fragments are analysed to establish how many fragments there are and which chromosomes they have come from. The laboratory counts the number of fragments from each chromosome of interest, and determines if there are more or less than expected. If, for example, there are more than expected number of fragments from chromosome 21, this indicates an increased chance of the baby having trisomy 21.

The results will be communicated from the test laboratory to East Lancashire Hospitals and communicated to you once this has been received. A result is normally available in 3-5 working days, but can sometimes be longer than this. If you do not hear anything back after 7 working days please contact Mr Maher's secretary or the screening team.

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NIPT is not diagnostic but a screening test. This means that the test has a certain sensitivity for each condition and can sometimes be wrong.

We discussed the sensitivity for Trisomy 21 is over 99% (false positive rate <0.1%)

Trisomy 18 is 97.4% (false positive rate <0.1%)

Trisomy 13 is 93.8% (false positive rate <0.1%)

I explained that the result will give a high or low risk result. If you wish to consider invasive/diagnostic tests following the results these can be arranged in the Fetal Medicine Unit.

Following counselling today, you wished to proceed with the test and written consent was obtained for this.

Ultrasound scan confirms a viable pregnancy.

Blood samples have been taken today using an aseptic non-touch technique and sent by recorded mail to the Harmony laboratory.

Follow-up

Results will normally be available in 3-5 working days and will be communicated to you via the screening team/Mr Maher.





porformed in the UK



The Halo Building 1 Mabledon Place London WC1H 9AX

T: +44 (0) 20 7307 7409 El xamelin Cidado (19) Can

www.idipathology.com

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DATE OF BIRTH: (OD/MAA/YYYY) MAN:		East Lancashire Hospitals Trust
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	4 wks 2 days	OTHER CLINICIAN:
# OF FETUSES:	IVF STATUS: non-IVF pregnancy	OTHER CLERICIANI CONTACT INFORMATION:
COLLECTION DATE (DD/A 10/04/2019	MI/YYY): RECEIVED DATE (DO/MM/YYY): 11/64/2019	REPORT DATE: [UDJ/AM/YYY) 15/04/2019

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CHROMOSOME	RESULT	PROBABILITY	RECOMMENDATION
Trisomy 21 (T21)	Low Probability	Less than 1/10,000 (0.01%)	Review results with patient
Trisomy 18 (T18)	Low Probability	Less than 1/10,000 (0.01%)	Review results with patiens
Trisomy 13 (T13)	Low Probability	Less then 1/10,000 (0.01%)	Review results with patient

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TEST DESCRIPTION

The Harmony Prenatal Test* measures the relative proportion of chromosomes to aid in the assessment of fetal trisomies 21, 18, and 13. Harmony's performs a directed analysis of cell-free DNA ICIDNA) in maternal blood and incorporates the fetal fraction of cfDNA in test results. Test results also incorporate maternal age (or egg donor age) and gestational age related probability based on information provided on the test requisition form. Probability of less than 1% is defined as low probability and 1% or greater is defined as high probability. Harmony has been validated in singleton and twin pregnancies of at least 10 weeks gestational age. Harmony is not validated for use in pregnancies with more than two fetuses, demised twin, mosaicism, partial chromosome aneuploidy, translocations, maternal aneuploidy, transplant, malignancy, or in women under the age of 18. Harmony does not detect neural tube defects. Twin results reflect the probability that the pregnancy. involves at least one affected fetus. Analysis of cIDNA does not always correlate with fetal genotype. Not all aneuploid fetuses will have a high probability result and some auploid fetuses will have a high probability result.

Fetal Sex test quantifies the Y chromosome. A "female" result indicates no detection of Y chromosome and a "male" result indicates detection of Y chromosome, it does not exclude sex chromosome arieuploidy. For twin pregnancies, a male result indicates one or two male letuses.

CUMICAL VALIDATION BATA FROM THE HARMONY PRODUCT INSERT*

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	Delection Raig	False Positive Rate
721	107/10R	0/641
718	25/25	0/641
713	11/12	0)641

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NHS Trust Lancashire Women & Newborn Centre Casterton Avenue Burnley BB10 2PQ

> Hospital No: NHS No.: Date 30/04/2019

Genetic Counselling

Patient:

ΙĘ

Exam date:

Indication

Raised risk on combined screening

Pregnancy

Singleton pregnancy, Number of fetuses: 1.

Test Results

Please find enclosed the results of your recent Harmony Non-invasive Prenatal Test (NIPT).

The results indicate that you are HIGH PROBABILITY for Trisomy 21 (Down Syndrome) and LOW PROBABILITY for Trisomy 18 (Edward Syndrome) and Trisomy 13 (Patau Syndrome).

The fetal sex was determined to be male.

The amount of fetal cells within the blood test was deemed sufficient to analyse the sample (fetal fraction 16.2%).

As you recall, we discussed that the NIPT is not a diagnostic test so will not detect all cases of chromosome problems. Only invasive tests (amniocentesis) are diagnostic at this stage. If you wish to proceed with an invasive test at this stage please contact the screening team.

The test will not detect mosaicism, partial chromosome problems, translocations or maternal chromosomal problems. Harmony does not detect structural problems with your baby - these are best detected at your 20-week anomaly scan.

Follow-up

In view of the raised probability result an appointment has been made for you with the screening team/Fetal Medicine Unit to discuss this further and discuss any further investigations that you may wish to consider.

If you wish to discuss the test any further in the meantime please feel free to contact my secretary on 01282-803197.

Consultant Obstetrician

CC:







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(DD/MM/OTY)

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GENETICS

The Halo Building 1 Mabledon Place London WC1H 9AX

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www.idipathology.com

LYTTER TRANS	<u> ខ្មែកពីវិសាធាតិ ដែល</u>
DATE OF BIRTH: (CO/MIA/YTY)	
MRII:	
LASORATORY ID:	OTHER ID:
GESTATIONAL AGE:	

13 wks 3 days

OF FETUSES: IVE STATUS: non-IVF pregnancy COLLECTION DATE (DD/AMA/YYYY) : RECEIVED DATE (DD/NAM/YYYY); 18/04/2019 23/04/2019

ACCOUNT # EASTLANC CUSIC HAME: East Lancashire Hospitals Trust ACTERNING/ORDERING CLINICIALS PEFERRITG/ORDERING CUMCUM EMAIL OTHER CLINICIAN CONTACT INFORMATION

29/04/2019

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CHROMOSOME	RESULT	PROBABILITY	RECOMMENDATION
irisoloya2 (jezi) .	High Fremability	Greaterthon SE/A00 (ESS)	Genetic courselling and additional resulting
Trisomy 18 (T18)	Low Probability	Less than 1/10,000 (0.01%)	Review results with patient
Trisomy 13 (T13)	Low Probability	Less than 1/10,000 (0,01%)	Deputeur Faculty (Disk Septemb

Fetal Sex Male
Fetal Sex Male
<u>L. N. S. C. C.</u>

TEST DESCRIPTION

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	Datection Asta	Faire Positive Rate
121	107/108	0/641
718	25/29	n/G41
713	12/12	0/641

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Lancashire Women & Newborn Centre Casterton Avenue Burnley BB10 2PQ

> Hospital No: NHS No.:

Genetic Counselling

Patient:

Exam date:

Indication

Pregnancy

Counselling Summary

Maternal request, Raised risk on combined screening

Singleton pregnancy. Number of fetuses: 1.

Attended for Non-invasive Prenatal Testing (NIPT) due to an increased chance result on the combined test for T21.

Explained that this is a private test not available on NHS at present. Harmony is the company of choice in ELHT, but other providers are available privately. Cost of the test is £350 (Harmony will invoice you for this directly).

Discussed that screening for fetal chromosomal disorders is optional. Implications for the test results discussed. The decision to proceed is based on how each individual perceives the benefits of obtaining the information about chromosomal problems when weighed against the potential emotional and physical risks of testing.

NIPT is a screening test. It cannot diagnose or eliminate the chance that a fetus has a particular chromosomal condition; rather it separates women into high and low risk categories. Testing is performed using a blood test of mother from the arm and is not associated with an increased risk of miscarriage.

We discussed that the only diagnostic test at this gestation is an amniocentesis test. This is a test where a small needle is placed through your abdomen under ultrasound guidance and a small amount of fluid surrounding the baby is taken for analysis. This will confirm if the baby is affected by a chromosomal condition. There is a small risk of 0.5-1% of miscarriage.

Clinical features of each condition screened for in the test discussed. These included:

- Trisomy 21 (Down Syndrome)
- Trisomy 18 (Edwards Syndrome)
- Trisomy 13 (Patau Syndrome)
- Sax chromosome aneuploidles (monosomy X, XXY, XXX, XYY). The test for this is optional. You decided that you 'DID NOT want to proceed with this test.
- Sex of the fetus. This test is optional. You have expressed that you wish this test to be performed.

Specific testing methods discussed — there are fragments of chromosomes that circulate outside of cells within the mothers blood. These DNA fragments are referred to as 'cell-free'. Most cell free DNA fragments derive from the mothers blood cells, but some come from the placenta. Because the fetus and placenta derive from the same fertilised cell, they are usually genetically identical. The sequences of cell free DNA fragments are analysed to establish how many fragments there are and which chromosomes they have come from. The laboratory counts the number of fragments from each chromosome of interest, and determines if there are more or less than expected. If, for example, there are more than expected number of fragments from chromosome 21, this indicates an increased chance of the baby having trisomy 21.

The results will be communicated from the test laboratory to East Lancashire Hospitals and communicated to you once this has been received. A result is normally available in 3-5 working days, but can sometimes be longer than this. If you do not hear anything back after 7 working days please contact Mr Maher's secretary or the screening team.

in 3 out of 100 women a repeat test will be required (a result is achieved in approximately two thirds of these). This is because the amount of fetal blood cells in your circulation is not enough for the laboratory to test. This is called the fetal fraction, if the test requires repeating you will not be charged for this.

NIPT is not diagnostic but a screening test. This means that the test has a certain sensitivity for each condition and can sometimes be wrong.

We discussed the sensitivity for Trisomy 21 is over 99% (false positive rate <0.1%)

Trisomy 18 is 97.4% (false positive rate <0.1%)

Trisomy 13 is 93.8% (false positive rate <0.1%)

I explained that the result will give a high or low risk result. If you wish to consider invasive/diagnostic tests following the results these can be arranged in the Fetal Medicine Unit.

Following counselling today, you wished to proceed, with the test and written consent was obtained for this.

Ultrasound scan confirms a viable pregnancy.

Blood samples have been taken today using an aseptic non-touch technique and sent by recorded mall to the Harmony laboratory.

Follow-up

Results will normally be available in 3-5 working days and will be communicated to you via the screening team/Mr Maher.

Consultant Obstetrician

Page 2 of 3 for report of patient