apple tree medical

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May 8, 2023

Private and Confidential

Dr. Nirjhar Nandi
Cairns Endocrinology
Suite 3G Flecker House
5 Upward Street
Cairns QLD 4870
Ph: 4051 9711
Fax: 4051 9707

Dear Dr. Nandi

Re: Ms Jennifer E Eustace - DOB: 25/03/89
3 Basalt Street
BRINSMEAD QLD 4870

Telephone home: 0433926992
mobile: 0478 089 013

Thank you for seeing Jennifer, a 34 year old lady previously reviewing with Cairns Hospital, who was hoping to transition to private, for review in regards to hypertension and recent PET and pregnancy loss.

Had been seeing obstetric medicine clinic who's letters I've attached. Was transferred to Townsville and unfortunately lost the pregnancy (letter attached). She was discharged on methyldopa and labetalol with control of her BP, though due to a shortage switched her to metoprolol, with last blood pressure 103/80, and 131/104 today though forgot to take her medication this morning.

I'd appreciate if you would be able to follow her up regarding her hypertension and whether you feel she requires any further secondary screening?

Current Problems:

- Severe Atopic Dermatits
- Hypertension
- Allergic rhinitis - onset 19/06/2019

Past History:

• Asthma

Medications:

• Aspirin 100mg Tablets 1 po daily

• Budamax Nasal spray 64mcg/dose Nasal spray 1 spray each nostril once daily

• Eltroxin Tablets 50mcg Tablets 50mg po 4 days per week, 75mg po 3 days per week

• Eltroxin Tablets 75mcg Tablets 50mg po 4 days per week, 75mg po 3 days per week

• Folic acid 5mg Tablets 1 po daily with food

• Methyldopa 250mg Tablets 1 po daily

• Metoprolol Sandoz Tablets 50mg Tablets 0.5 po BD

• Temazepam 10mg Tablets 0.5-1 po nocte prn

Allergies:

• Nil Known

Kind regards

Dr Michael Breitkreutz
Provider No:4910827L

Diagnostic Results: Ms Jennifer E Eustace
Result IDs: (309815 309816 309817 309818 309819 309821 309822 309823 309837 309838 309850 309855 310074 310098 310246 310467 310529 310603 310968 311884 312379 312746 313161 314696 314981 316654 316893 316910)

E/LFT ()
Collection Date: 11:15 13/04/2023
CUMULATIVE SERUM BIOCHEMISTRY

Date	23/03/22	01/04/23	13/04/23		
Time	09:36	12:15	11:15		
Lab No	69472786	72277976	72891304		
	RANDOM	RANDOM	RANDOM		RANDOM
Sodium	140	139	141	mmol/L	(137-147)
Potass.	4.0	4.2	4.6	mmol/L	(3.5-5.0)
Chloride	104	105	102	mmol/L	(96-109)
Bicarb	28	25	27	mmol/L	(25-33)
An.Gap	12	13	17	mmol/L	(4-17)
Gluc	4.7	4.0	5.0	mmol/L	(3.0-7.7)
Urea	4.1	5.0	5.8	mmol/L	(2.0-7.0)
Creat	51	52	66	umol/L	(40-110)
eGFR	> 90	> 90	> 90	mL/min	(over 59)
Urate	0.33	0.36	0.51	mmol/L	(0.14-0.35)
T.Bili	7	10	4	umol/L	(2-20)
Alk.P	121	115	104	U/L	(30-115)
GGT	29	32	38	U/L	(0-45)
ALT	101	208	37	U/L	(0-45)
AST	48	110	18	U/L	(0-41)

LD	334	522	203	U/L	(80-250)
Calcium	2.42	2.26	2.35	mmol/L	(2.15-2.60)
Corr.Ca	2.35	2.45	2.36	mmol/L	(2.15-2.60)
Phos	1.1	1.2	1.1	mmol/L	(0.8-1.5)
T.Prot	74	60	70	g/L	(60-82)
Alb	45	36	42	g/L	(35-50)
Glob	29	24	28	g/L	(20-40)
Chol	4.6		6.8	mmol/L	(3.6-6.7)
Trig	1.4		2.3	mmol/L	(0.3-4.0)
Lab No	69472786	72277976	72891304		
Date	23/03/22	01/04/23	13/04/23		

Tests Completed:FBC, SE E/LFT
Tests Pending :

FULL BLOOD COUNT
Collection Date: 11:15 13/04/2023
CUMULATIVE FULL BLOOD EXAMINATION

Date	23/03/22	03/12/22	01/04/23	13/04/23		
Time	09:36	08:40	12:15	11:15		
Lab No	69472786	71974230	72277976	72891304		
Hb	150	142	111	127	g/L	(115-160)
RCC	5.2	4.9	3.9	4.4	x10 ^12 /L	(3.6-5.2)
Hct	0.45	0.45	0.33	0.41		(0.33-0.46)
MCV	86	91	84	95	fL	(80-98)
MCH	29	29	29	29	pg	(27-35)
Plats	360	367	82	537	x10 ^9 /L	(150-450)
WCC	8.5	7.5	10.7	10.2	x10 ^9 /L	(4.0-11.0)
Neuts	4.4	4.4	7.8	67 % 6.8	x10 ^9 /L	(2.0-7.5)
Lymphs	2.4	1.9	1.5	23 % 2.3	x10 ^9 /L	(1.1-4.0)
Monos	0.5	0.5	0.9	6 % 0.6	x10 ^9 /L	(0.2-1.0)
Eos	1.19	0.68	0.54	4 % 0.41	x10 ^9 /L	(0.04-0.40)
Basos	0.00	0.08	0.00	0 % 0.00	x10 ^9 /L	(< 0.21)

72891304 Automated Comment:
As per ISLH guidelines - Film not reviewed. If a film review is truly indicated, contact the laboratory within 24 hours of collection. Otherwise investigate any highlighted abnormalities as clinically appropriate.

Eosinophilia - this may be seen in Atopic conditions. Correlate Clinically. Consider IgE +/- RAST.

Thrombocytosis may be seen in hyposplenic states, or occur as response to infection, inflammation, post-surgery/trauma, malignancy, bleeding, or in older patients an early Myeloproliferative Neoplasm (MPN). Correlate with E+LFTs, ESR/CRP, Iron Studies. If a MPN is a possibility, JAK2 molecular analysis is recommended.

** FINAL REPORT - Please destroy previous report **

Tests Completed:FBC
Tests Pending :SE E/LFT

QH Patient correspondence
Collection Date: 14:18 05/04/2023
[]OBSTETRIC MEDICINE CLINIC

Date: 20 March 2023

Ref: ZG/bk UR: 633955

Fin: 3355780

Dr Michael Breitkreutz

Apple Tree Medical

PO Box 712N

CAIRNS NORTH QLD 4870

Dear Dr Breitkreutz

RE: JENNIFER EUSTACE D.O.B. 258/03/1989

Your patient, as above, was reviewed in Clinic today. Following are my notes regarding this.

33yo F
G1P0
24 weeks
Issues:
Chronic HTN
- element of white coat HTN
- initially raised plasma metanephrines, normal on repeat
- raised salivary cortisol of unclear significance
- recent admission under O&G, labetalol 200mg TDS added (already on methyldopa 250mg BD)
Hypothyroidism
- antibody status unknown
- TSH in target: 7/1: TSH 1.8, FT4 14
- current dosing: thyroxine 50mcg daily with 75mcg every 3rd day
Risk of PET
- recommended LDA
Early OGTT
- negative
Severe atopic dermatitis
- known to Derm
- was on dupilumab prior to pregnancy
- has previously been on azathioprine and ciclosporin
- was on long-term prednisolone until 4 years ago
- reports dermatitis clearing to the point that she was able to cease treatment after acupuncture (which she gets every 2 weeks)
Asthma
- never been hospitalised
- not required treatment for years

Investigations:
Plasma metanephrines (QML) 20/2/23 - all WNL
3-methoxytyramine <35
Normetadrenaline 479
Metadrenaline <75

Previous plasma metanephrines (QML) 9/12/22

3-methoxytyramine <35
Normetadrenaline 625
Metadrenaline 88
Salivary cortisol 10/2/23 (QML)
0700hrs 5.6nmol/L
1400hrs 4.6nmol/L
2400hrs 5.3nmol/L
Review:
Feeling well since previous discharge
No headache/visual disturbance
No N/V
No RUQ pain
Some chronic swelling to ankles - denies recent change
Asthma well controlled, no flares
No flares of dermatitis
States has been checking BP at home and at home
- systolic 120-135
- diastolic 80-95
Has not been documenting BPs
States gets significant anxiety re coming to appointments and this puts up her BP
Was not keen to stay in hospital 2 weeks ago when she was admitted for hypertension
Initial BP check = 164/124
Repeat measurement 164/92 (manual)
Chest clear
No RUQ/epigastric tenderness
No hyperreflexia, 1 beat clonus bilat
No significant oedema
Imp:
Suboptimal BP control with likely element of white coat hypertension exacerbating this during appointments
Long discussion with patient

- offered admission for serial BP monitoring, extra dose of labetalol - could aim to discharge home if BP normalised

- patient anxious and upset, does not feel that remaining in hospital will allow BP to normalise

- does not want to stay in hospital overnight

- long discussion with patient re risks of uncontrolled hypertension in pregnancy, concerns re pre-eclampsia

- pt understanding of risks and our concerns, able to repeat these back to me

- declines to stay in hospital, feels BP will be better controlled at home in her own environment

- has supportive partner at home

- will recheck BP tonight and if persistently or severely elevated will represent

- planning to re-present for O&G appointment tomorrow morning

Plan:
Offered admission for BP monitoring and optimisation - pt has declined, aware of risks (see above)
Increase methyldopa to 500mg BD
Continue labetalol dose as is
Encouraged patient to monitor BP multiple times/day at home and document these - bring to next appointment
Written information provided to patient re BP targets and when to re-present
Review with O&G tomorrow morning
Form for PET screen to do prior to next appointment

Obs Med OPD 2/52
Will need repeat TFTs 3rd trimester - have not provided form for this yet
Above d/w Dr Nandi

Yours sincerely

Electronically verified

ZOE GAVEY

OBSTETRIC MEDICINE REGISTRAR

ULTRASOUND RECALL FOR FURTHER IMAGES
Collection Date: 00:00 07/03/2023

Patient ID: 74.CCE667Z
Dr Michael Breitkreutz Order: 77.42608184_1

Dr Michael Breitkreutz
Apple Tree Medical Cairns
447 - 449 Draper St
Parramatta Park 4870
Tel: 0740511074
7th March 2023

Patient ID: 74.CCE667Z
Accession Number: 77.42608184
Reported: 9 March 2023

Dear Dr Breitkreutz

Re: Ms Jennifer Eustace - DOB: 25/03/1989
3 Basalt St BRINSMEAD 4870

History:

Week ago patient complained morphology scan.

Findings:

Today's imaging was again very difficult due to maternal and foetal factors.

The brain and facial images were obtained and appear grossly normal. Fetal heart
and corpus callosal views were unable to be obtained.

Fetal heart rate 153 BPM.
Cervix long and closed.
Placenta 3.6 cm from the internal os.

Conclusion:

Fetal anatomy scan remains incomplete with a foetal heart and corpus callosum
views unable to be obtained.

Consider an early 3rd trimester growth scan with repeated attempt to assess
these structures.

Dr Daniel Webb
Electronically signed at 2:28 pm Thu, 9th Mar 2023

US OBSTETRIC 17-22 WKS (1ST PRESENTATION)
Collection Date: 00:00 22/02/2023

Patient ID: 74.CCE667Z
Dr Michael Breitkreutz Order: 77.42404984_1

Dr Michael Breitkreutz
Apple Tree Medical Cairns
447 - 449 Draper St
Parramatta Park 4870
Tel: 0740511074
Patient ID: 74.CCE667Z
Accession Number: 77.42404984
22nd February 2023 Reported: 22 February 2023

Dear Dr Breitkreutz

Re: Ms Jennifer Eustace - DOB: 25/03/1989
3 Basalt St BRINSMEAD 4870

Ultrasound obstetrics:

Clinical Indications:

Morphology scan

Findings:

Anatomy Scan
Fetal cardiac activity: Present. 148bpm.

Fetal measurements:
Biparietal diameter (BPD) 46mm. 33%.
Head circumference (HC) 176mm. 34%.
Abdominal circumference (AC) 148mm. 38%.
Femur length (FL) 32mm. 31%.
Humerus length (HL) 31mm.
Estimated Fetal Weight (EFW) 329g. 33%.

Fetal anatomy:
The following were visualised and appeared normal for gestational age: cranium,
diaphragm, stomach, kidneys, bladder, umbilical cord, upper limbs, lower limbs,
hands, feet and spine.
The following could not be adequately assessed today: 4 chambers, interventricular septum, transverse arch, LVOT, RVOT, ductal arch, ventricles,
cisterna magna, cerebellum, nuchal fold, nasal bone, mandible, orbits/ lenses,
nose/lips and nasal bone/profile.
Placental cord insertion 2cm from superior margin of placenta.

Amniotic fluid: Normal.
Placenta: Anterior and 36mm from os.

Cervix: closed measuring 48mm.
No adnexal abnormality.

Conclusion:

Single intrauterine pregnancy.
Gestational age : 20 weeks 2 days.
EDD : 10/07/2023.
Placental cord insertion 2cm from superior margin of placenta, borderline
marginal insertion.
Incomplete anatomy scan. Visualisation of the fetal heart, brain, profile and
facial structures were limited by fetal position. These will be completed at a
subsequent scan on 7/3/23. No obvious abnormality seen today.

Signer Name

Dr Ahmed Kamalaldin
Electronically signed at 1:25 pm Wed, 22nd Feb 2023

cc: Dr Cairns Base Hospital

cc: CAIRNS BASE HOSPITAL, Dr Antenatal Clinic, Cairns Base Hospital Antenatal Clinic CAIRNS QLD 4870

QH Patient correspondence
Collection Date: 13:55 10/02/2023
[]OBSTETRIC MEDICINE CLINIC

Date: 19 January 2023

Ref: PH/bk UR: 633955

Fin: 3295128

Dr Michael Breitkreutz

Apple Tree Medical

PO Box 712N

CAIRNS NORTH QLD 4870

Dear Dr Breitkreutz

RE: JENNIFER EUSTACE D.O.B. 25/03/1989

Your patient, as above, was reviewed in Clinic today. Following are my notes regarding this.

Presenting Complaint

33 yo F P0 K15, EDD 10/7/23
New, referred for chronic HTN with abnormal secondary HTN screen
Issues:
Chronic HTN
- element of white coat HTN
- mildly raised metanephrines and salivary cortisol of uncertain significance
- awaiting renal tract ultrasound and 24 hour BP monitoring
- needs TTE
Hypothyroidism
- antibody status unknown
- TSH in target: 7/1: TSH 1.8, FT4 14
- uncertain of current thyroxine dose
Risk of PET
- has been recommended to take aspirin but not yet taking
Early OGTT
- negative
Severe atopic dermatitis
- known to Derm
- was on dupilumab prior to pregnancy
- has previously been on azathioprine and ciclosporin
- was on long-term prednisolone until 4 years ago
- reports dermatitis clearing to the point that she was able to cease treatment after acupuncture (which she gets every 2 weeks)
Asthma
- never been hospitalised
- not required treatment for years
Social History
- social worker
- non-smoker and no alcohol in pregnancy. No illicit drugs.

- lives with partner
- regularly exercises

RE: JENNIFER EUSTACE D.O.B. 25/03/1989 ??Con?t

History of Present Illness
Discussed the diagnosis of likely chronic HTN, despite the element of white coat HTN. Takes her BP at work once a day and typically has readings in the high 130s/90s.
Discussed the risk of developing PET with her history of HTN, what to monitor for and when to present. Her GP had suggested that she start taking low dose aspirin, however she has not yet been taking it - reinforced that this will decrease the likelihood of developing PET, along with adequate dietary calcium. Also explored the option of Jen buying her own BP monitor to take BP twice per day. Discussed that some of her secondary hypertension screen was mildly abnormal and that this is of uncertain significance while pregnancy but requires repeating and additional testing.
No history of fragility fractures, striae, DM or cushingoid feature. Did not note central adiposity prior to pregnancy. Noted peripheral oedema. Jen said this normal for her. No history of orthopnoea or dyspnoea. No chest pain or palpitations.
Discussed TFT results - TSH within target (1.8 on 7/1).
BP initially elevated during consultation - however reports she was very anxious about having it taken. Initial BP 163/100 -> repeated 10 minutes later and it was 140/95. Asymptomatic. Discussed the plan to commence methyldopa (nb. nation-wide shortage of labetalol), however it can impact on the tests we want to do, so asked to repeat them prior to commencing the medication. Jen is going away next week and was eager to start the medications, but we discussed that the treatment wasn't urgent at this stage, so ok to delay commencing medications until after then and the tests were complete. No history of depression.
Physical Exam
Observations & Measurements
Alert and lucid
Appeared somewhat overwhelmed during the consultation.
HSDNM
Lungs clear
Woody peripheral oedema - non-pitting and appeared chronic. Jen states this is normal for her.
JVPNE (sitting upright)
Initial BP 163/100 -> repeated 10 minutes later and it was 140/92

Assessment and Management Plan

D/w Dr Li with thanks.
Plan:
- repeat salivary cortisol (3 samples, at 7 am, 2pm and midnight)
- 24 hour urine cortisol
- awaiting renal artery doppler
- Referred for TTE (HeartRx)
- baseline urine PCR
- No change to thyroxine dose; repeat in third trimester
- Advised to take aspirin 100mg nocte from now
- prescribed methyldopa 250mg BD, however asked not to take it until the above tests have been done as it can affect results (not urgent to start)
- phone review in 4 weeks
- advised to purchase her own BP monitor and check BD: present if BP > 160, or see GP if BP frequently > 140/150.
- given verbal and written advice about PET and when to seek medical review
- needs regular BP checks with midwife/GP/at hospital

Yours sincerely

Electronically verified

PRUDENCE HOGG

REGISTRAR

THYROID TEST
Collection Date: 12:20 04/02/2023
CUMULATIVE SERUM THYROID FUNCTION TESTS

Date	23/03/22	03/12/22	07/01/23	04/02/23	
Time	09:36	08:40	10:09	12:20	
Lab No	69472786	71974230	70941488	71619776	
TSH	3.3	2.8	1.8	2.0	mIU/L (0.50-4.00)
free T4	15	14	14		pmol/L (10-20)

The TFT reference intervals for pregnancy are as follows:

	TSH (mIU/L)	FT4 (pmol/L)
First trimester	0.03-2.5	10-21
Second trimester	0.05-3.0	11-18
Third trimester	0.30-3.5	9-17

Progress level - On Levothyroxine
The therapeutic target for replacement therapy in primary hypothyroidism is to maintain a normal TSH but it is reasonable to aim for a TSH level in the lower reference interval if the patient has persistent symptoms.
In patients with differentiated thyroid cancer, the target TSH level should be subnormal or undetectable depending on individualized post-ablation risk stratification.
Note: Serum TSH should not be used to monitor T4 therapy in central hypothyroidism. Serum fT4 level should be maintained in the upper 50% of the normal range.

Tests Completed:TFT
Tests Pending :

NIPT Report
Collection Date: 09:01 14/01/2023

Sullivan Nicolaides PATHOLOGY

Jennifer Eustace

DOB	25 Mar 1989 (33 Yrs)
Lab ID	669662640
Phone	0478089013
Address	3 Basalt Street BRINSMEAD QLD 4870
Your Ref	
Ref by	Dr M Breitkreutz (B14936)
Requested	14 Jan 2023
Collection	14 Jan 2023, 09:01
Reported	19 Jan 2023, 11:05

Dr Michael R Breitkreutz
Apple Tree Medical
447 Draper St
CAIRNS QLD 4870

B14936
CRND,CRN3/---/---/---/---

Non-invasive prenatal test (NIPT) report

Indication: antenatal screening test for fetal aneuploidy

SAMPLE TYPE	ESTIMATED FETAL FRACTION	GESTATION	NO. OF FETUSES
Maternal plasma (cfDNA)	3% (sufficient for analysis)	14 weeks 5 days (at collection)	1

TEST	RESULT	RECOMMENDATION
Trisomy 21 (T21)	Low probability	Review results with patient
Trisomy 18 (T18)	Low probability	Review results with patient
Trisomy 13 (T13)	Low probability	Review results with patient
Fetal sex	Male	Review results with patient
Sex chromosome aneuploidy (SCA)	Not requested	

INTERPRETATION

Testing of placental cell-free DNA from this sample showed no evidence of the chromosomal abnormalities tested. "False negative" NIPT results are rare and can occur due to biological or analytical factors. Patients with a high pre-test probability of an abnormality will have a higher residual risk after NIPT than those with a low pre-test probability. Screening for fetal sex involves detection of Y chromosome material. The presence of Y chromosome material is interpreted as "male sex"; the absence of Y chromosome material is interpreted as "female sex". The result for fetal sex does not exclude sex chromosome aneuploidy.

NIPT IS A SCREENING TEST

Non-invasive prenatal testing (NIPT) measures the relative abundance of cell-free DNA (cfDNA) from various chromosomes in a sample of maternal blood collected at 10 or more weeks gestation. The test screens for selected chromosome disorders and (optionally) sex in the fetus and placenta. NIPT is a screening test for an uncommon event. The chance that a report of "low probability" in the fetus is correct (i.e. true negative) will vary with the pre-test probability of that disorder. The accuracy of this test is dependent on the provided gestational age and parity (as shown above); please contact the laboratory immediately if this information is incorrect. Please refer to p2 of this report for further test information.

The VeriSeq NIPT Solution v2 test was performed at Sullivan Nicolaides Pathology

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24 Hurworth Street, Bowen Hills Qld 4006
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IS12APR2022

Sonic Genetics

NATA Accreditation No. 1964
RCPA The Royal College of Pathologists of Australasia
Accredited for compliance with NPAAC standards and ISO 15189

Sullivan Nicolaides PATHOLOGY

Jennifer Eustace

Lab ID 669662640 **DOB** 25 Mar 1989 (33 yrs)
Referring Doctor Dr M Breitkreutz (B14936)
Reported 19 Jan 2023, 11:05

SCOPE OF NIPT

NIPT is a screening test for specific chromosome disorders as selected on the request form and identified in the report. NIPT does not screen for other chromosome disorders or balanced translocations. NIPT is not a test for structural abnormalities of the fetus such as neural tube defects.

The determination of fetal sex by NIPT is not a test for medical purposes and is not the primary intent of NIPT. If sufficient Y chromosome cfDNA is detected, the reported sex is 'male'; otherwise, the reported sex is 'female'.

The report for twins reflects the probability that at least one twin has the specified disorder. In relation to fetal sex, a male result indicates at least one male twin.

This NIPT has been validated in singleton and twin pregnancies at 10 or more weeks gestation. The results of the test can be confounded and invalidated by certain maternal and fetal factors including but not limited to the following; pregnancies with >2 fetuses, demised twin, fetoplacental mosaicism, maternal mosaicism, maternal aneuploidy, organ transplant or malignancy.

CONFIRMATION OF RESULTS

Fetoplacental cfDNA is largely derived from the placenta. The chromosome status of the placenta is usually identical to that of the fetus. However, if in the rare event that they are not, NIPT may provide a result which differs from the true status of the fetus. This reflects the complex biology of pregnancy. As a result, on average, up to 1 in 1,000 fetuses with normal chromosomes will have a false positive NIPT result for one of the common trisomies, while up to 1 in 100 fetuses with one of the common trisomies will have a false negative NIPT result. False positive and negative results for sex chromosome disorders, rare autosomal aneuploidies and partial deletions/duplications may be more frequent (see NIPT performance table).

For this reason, NIPT is not a diagnostic test of the fetus. The result should be considered together with other information, and confirmatory testing should be done prior to decision-making. The table at right documents the performance of NIPT when assessed against the chromosome status of the fetus.

POSITIVE AND NEGATIVE PREDICTIVE VALUES

The PPV and NPV of a test for a specific chromosome abnormality depend on the sensitivity, specificity, and pre-test probability that the pregnancy has a fetus with that abnormality. The pre-test probability can vary widely depending on factors not known to the laboratory. For that reason, we cannot provide the PPV or NPV for a given patient. Assuming a sensitivity and 1-specificity of NIPT for a specific abnormality of 99% and 0.1% respectively, then the estimated PPV and NPV for different pre-test probabilities are shown below:

Pre-test probability	1 in 10	1 in 100	1 in 1,000
PPV	99.1%	90.9%	49.8%
NPV	99.9%	99.99%	>99.99%

QUALITY METRICS

At 12 weeks' gestation, an average of 10% of the cfDNA in maternal plasma is derived from the fetus/placenta, with the observed range being approximately 1 - 30%. As part of the NIPT assay, each sample is tested to determine whether the fetoplacental DNA quantity and quality metrics are sufficient to provide a reliable result.

NIPT performance

This sample was tested using VeriSeq NIPT Solution v2, a TGA-registered NIPT, which is supplied by Illumina Australia.

The following information is derived from information provided by the manufacturer of this test. The performance for detecting trisomies 21, 18 and 13 was derived from outcomes in 2,236 singleton pregnancies. The performance for detecting monosomy X, XXX, XXY and XYY was derived from outcomes in singleton pregnancies; screening for sex chromosome aneuploidy is not available for twins.

The concordance between predicted fetal sex and genital phenotype of the newborn was >99.9% (95% CI 99.8-100%).

Note that given statistical sample size and biological limitations, not all aneuploid fetuses will be classified as high probability, and some disomic fetuses will be classified as high probability.

Chromosome status in fetus	Sensitivity (95% CI)*	1-specificity (95% CI)#
Trisomy 21 (n=130)	>99% (97.1-100%)	0.10% (0.03-0.37%)
Trisomy 18 (n=41)	>99% (91.4-100%)	0.10% (0.03-0.36%)
Trisomy 13 (n=26)	>99% (87.1-100%)	0.10% (0.03-0.36%)
Sex chromosome aneuploidy (n=73)	95.9% (88.6-98.6%)	0.15% (0.05-0.44%)
Rare autosomal aneuploidy (n=28)	96.4% (82.3-99.4%)	0.20% (0.08-0.51%)
Partial deletions duplications >7Mb (n=27)^	74.1% (55.3-86.8%)	0.20% (0.08-0.51%)

* Proportion of pregnancies with a fetus or placenta having the abnormality correctly identified as 'high probability' by the test (trisomies) or of pregnancies with concordant results by NIPT and karyotype (sex chromosome aneuploidies).

\# Proportion of pregnancies not having the abnormality incorrectly identified as 'high probability' of the abnormality by the test; equivalent to false positive rate.

^ Sensitivity estimate for partial deletions/duplications is on a per-patient rather than per-anomaly basis. Some fetuses have more than one partial deletion/duplication >7Mb.

In a simulation study of twin pregnancies, the estimated sensitivities for detecting trisomies 21, 18 and 13 were 96.4%, 95.7% and 93.6% respectively.

Reference
Grati FR, Malvestiti F, Ferreira JCPB, Bajaj K, Gaetani E, et al. Genetics in Medicine 16 (2014): 620-624.

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Sonic Genetics

NATA Accreditation No. 1964
RCPA The Royal College of Pathologists of Australasia
Accredited for compliance with NPAAC standards and ISO 15189

FIRST TRIMESTER SCREEN
Collection Date: 08:00 31/12/2022
FIRST TRIMESTER DOWN'S SCREEN

Sample Taken 31/12/2022
Sample Number 22-70941155

Manufacturer: BRAHMS KRYPTOR
Analysed on 04/01/23

free beta HCG 26 IU/L Lot No B09136

PAPP-A 3.0 IU/L Lot No 766138

INTERIM REPORT - Pending measurement of Nuchal Translucency by Ultrasound, and final reporting of Overall Risk. This report is for information only -- for incorporation into the Ultrasound assessment.

Tests Completed:FIRST TRIMESTER SCREEN
Tests Pending :

US OBSTETRIC - NUCHAL LUCENCY
Collection Date: 00:00 30/12/2022

Patient ID: 74.CCE667Z
Dr Michael Breitkreutz Order: 77.41819721_1

Dr Michael Breitkreutz
Apple Tree Medical Cairns
447 - 449 Draper St
Parramatta Park 4870
Tel: 0740511074

Patient ID: 74.CCE667Z
Accession Number: 77.41819721

30th December 2022 Reported: 5 January 2023

Dear Dr Breitkreutz

Re: Ms Jennifer Eustace - DOB: 25/03/1989
3 Basalt St BRINSMEAD 4870

OBSTETRIC SCAN
Clinical Indications:Routine nuchal translucency

Gestational age by earliest ultrasound: 12 weeks 4 days.
Findings:
Suboptimal scan due to maternal body habitus
First Trimester Ultrasound
Both transabdominal and transvaginal scans were performed.
Fetal cardiac activity: Present.
Fetal heart rate: 174 bpm

Fetal measurements:
Crown-rump length (CRL): 70mm.
Nuchal translucency (NT): 1.3mm (prone TV images)
Nasal bone: Not visualised due to technical factors.

Fetal anatomy:
The following were visualised and appeared normal for gestational age: skull, brain, bladder, abdominal wall, spine, stomach, upper limbs and lower limbs.

Risk assessment
The maternal age-related risk for trisomy 21 is 1:361.
This results in a risk of 1:7224 using the above nuchal translucency and hormone measurements.
The maternal age-related risk for trisomy 18 is 1:907.
This results in a risk of 1:18149 using the above nuchal translucency and hormone measurements.
The maternal age-related risk for trisomy 13 is 1:2840.
This results in a risk of 1:3993 using the above nuchal translucency and hormone

measurements.
Placenta: Developing anteriorly.
Amniotic fluid: Normal.
Cervix: closed measuring 45mm.
The serum hormone levels are:
Free B-hCG = 1.108 MoM
PAPP-A = 1.199 MoM

Conclusion:
Single live intrauterine gestation corresponding to 13 weeks 1 day
Combined 1st trimester screening, low risk
Adequate cervical length
Established EDD :10/07/2023.

Please note this is a suboptimal scan for the assessment for NT and also for the
fetal anatomy. NB could not be assessed
An option of NIPT can be offered to the patient for trisomy screening.

Dr Jeyprada Deenadayaalu
Electronically signed at 4:42 pm Thu, 5th Jan 2023

METANEPHRINE,URINE
Collection Date: 06:49 05/12/2022
URINE CATECHOLAMINE STUDIES(NON-24 HOUR)

Metanephrine	0.2	umol/L	24 h(0.2-1.5)
Normetanephrine	1.5	umol/L	24 h(0.3-2.0)
3-Methoxytyramine	0.6	umol/L	
3-MT/Creatinine ratio	103	umol/mol	(< 197)
Creatinine	5.8	mmol/L	24 h(5.0-15.0)

Please note - Reference Interval for 24-hour urinary dopamine has been revised for patients older than 16 years.

Tests Completed:RENIN/ALDOSTERONE RATIO, SALIVA CORTISOL
Tests Pending :

Misc Chemistry
Collection Date: 06:49 05/12/2022

R.RANGE

Biochem. Misc 1 Salivary Cortisol
Chem. Misc. Salivary Cortisol (LC-MS/MS): 5.5 nmol/L

Salivary Cortisol Reference Intervals:
Cortisol AM (09:00) 3 - 35 nmol/L
Cortisol PM (16:00) 0 - 12 nmol/L
Cortisol MID (23:00) 0 - 5 nmol/L

CORTISOL, SALIVA
Collection Date: 06:49 05/12/2022

Salivary Cortisol (LC-MS/MS)	5.5	nmol/L

Reference Intervals:

Cortisol AM (09:00)	3-35 nmol/L
Cortisol PM (16:00)	0-12 nmol/L
Cortisol MID (23:00)	0-5 nmol/L

The Salivary Cortisol (LC-MS/MS) assay was performed and reported by Royal Brisbane Hospital (QHPS), QLD.

Tests Completed:RENIN/ALDOSTERONE RATIO, SALIVA CORTISOL
Tests Pending :

RENIN/ALDOSTERONE RATIO
Collection Date: 06:49 05/12/2022

	Aldosterone	370	pmol/L	Upright	(100-950)
+	Renin	78	mU/L	Upright	(3-40)
	Aldosterone/Renin Ratio	5			(2-75)

Ranges assume normal salt intake and Potassium levels, and not on antihypertensives or diuretics.

High Renin, normal Aldosterone : not suggestive of a primary Renin excess - ?Secondary to Antihypertensives. Not suggestive of HyperAldosterone syndromes.

Tests Completed:RENIN/ALDOSTERONE RATIO
Tests Pending :SALIVA CORTISOL

METADRENALINES,PLASMA
Collection Date: 06:49 05/12/2022
PLASMA METADRENALINES

	3-Methoxytyramine	< 35	pmol/L	(< 90)
+	Normetadrenaline (free)	625	pmol/L	(< 600)
	Metadrenaline (free)	88	pmol/L	(< 450)

The reported reference intervals are for recumbent patients.

Provided the patient was seated for collection, the plasma normetadrenaline result is within the seated reference interval for age (up to 970 pmol/L). This does not suggest phaeochromocytoma.

Dr George Marshall

Tests Completed:
Tests Pending :RENIN/ALDOSTERONE RATIO, SALIVA CORTISOL

CORTISOL, SALIVA
Collection Date: 06:49 05/12/2022
SALIVARY CORTISOL

This sample has been referred to the Royal Brisbane Hospital (QHPS) and results are expected within 3 weeks.

Tests Completed:
Tests Pending :RENIN/ALDOSTERONE RATIO, SALIVA CORTISOL

QUANTITATIVE HCG, SERUM
Collection Date: 08:40 03/12/2022

CUMULATIVE SERUM HUMAN CHORIONIC GONADOTROPHIN

Date	03/12/22
Time	08:40

Lab No 71974230

HCG 130000 IU/L

Serum HCG Test Interpretation
Non-pregnant: <5 IU/L (if taken after first missed period)
Borderline result: 5-25 IU/L (suggest repeat in 48 hours)
Pregnant: >25 IU/L
Post menopause: <10 IU/L

Ranges by Week : lower - Median - upper

4	40	350	2000
5	400	1800	10000
6	1600	8000	33000
7	7000	25000	65000
8	20000	50000	100000
9	40000	65000	120000
10	46000	70000	125000

Note - HCG levels peak at 10-12 weeks gestation and fall thereafter.

Tests Completed:HEP B / C, TFT, SYPHILIS AB, RUBELLA IGG, IRON STUDIES, CT NG PCR, FBC
Tests Completed:URINE MCS, GTT, GROUP ANTIBODY AND HOLD, HCG(QUANT), HIV 1/2 AB
Tests Pending :

THYROID TEST
Collection Date: 08:40 03/12/2022
CUMULATIVE SERUM THYROID FUNCTION TESTS

Date	06/03/17	23/03/22	03/12/22	
Time	12:17	09:36	08:40	
Lab No	64485114	69472786	71974230	
TSH	3.5	3.3	2.8	mIU/L (0.50-4.00)
free T4			15	pmol/L (10-20)

The TFT reference intervals for pregnancy are as follows:

	TSH (mIU/L)	FT4 (pmol/L)
First trimester	0.03-2.5	10-21
Second trimester	0.05-3.0	11-18
Third trimester	0.30-3.5	9-17

Euthyroid level.

Tests Completed:HEP B / C, TFT, SYPHILIS AB, RUBELLA IGG, IRON STUDIES, CT NG PCR, FBC
Tests Completed:URINE MCS, GTT, GROUP ANTIBODY AND HOLD, HIV 1/2 AB
Tests Pending :HCG(QUANT)

CTNG TV MG
Collection Date: 08:40 03/12/2022

Specimen: Urine

CHLAMYDIA TRACHOMATIS (NAT) Negative
(Nucleic acid Amplification)

NEISSERIA GONORRHOEAE (NAT) Negative
(Nucleic acid Amplification)

The Roche Cobas 6800 Nucleic Acid Amplification assay has been performed on this specimen.

Tests Completed:HEP B / C, SYPHILIS AB, RUBELLA IGG, IRON STUDIES, CT NG PCR, FBC
Tests Completed:URINE MCS, GTT, GROUP ANTIBODY AND HOLD, HIV 1/2 AB
Tests Pending :TFT, HCG(QUANT)

RUBELLA VIRUS
Collection Date: 08:40 03/12/2022

MICROBIAL SEROLOGY

Rubella IgG (EIA): POSITIVE

Negative Rubella IgG range : < 10 IU/mL
Low Positive Rubella IgG range : 10 to 29 IU/mL
Positive Rubella IgG range : >= 30 IU/mL

IMMUNE
This test determines immune status ONLY. If current/recent infection
is suspected, Rubella IgM testing is indicated.

Tests Completed:HEP B / C, SYPHILIS AB, RUBELLA IGG, IRON STUDIES, FBC, URINE MCS, GTT
Tests Completed:GROUP ANTIBODY AND HOLD, HIV 1/2 AB
Tests Pending :TFT, CT NG PCR, HCG(QUANT)

GROUP ANTIBODY AND HOLD
Collection Date: 08:40 03/12/2022

BLOOD GROUP and ANTIBODY SCREEN

Blood Group	O Rh(D) Positive
Red Cell Antibodies	Not Detected

Ordering of blood products for COVID positive patients should be focused and limited to 1 unit ordering per standard transfusion. Any blood products delivered to the bedside of a Covid positive patient CANNOT be returned to the hospital blood fridge and will be discarded.

Please contact your local pathology to organise collection of unused
products.

Tests Completed:HEP B / C, SYPHILIS AB, IRON STUDIES, FBC, URINE MCS, GTT
Tests Completed:GROUP ANTIBODY AND HOLD, HIV 1/2 AB
Tests Pending :TFT, RUBELLA IGG, CT NG PCR, HCG(QUANT)

URINE MICRO / CULTURE
Collection Date: 08:40 03/12/2022

EXAMINATION OF URINE

Collection:	Mid stream urine
Protein:	+
Glucose:	Negative
Ketones:	Negative

PHASE CONTRAST MICROSCOPY

Leucocytes:	30	x10 ^6 /L	(N.R. <10)
Erythrocytes:	10	x10 ^6 /L	(N.R. <10)
Epithelial:	80	x10 ^6 /L	

CULTURE

No significant growth after overnight incubation.

Tests Completed:HEP B / C, SYPHILIS AB, IRON STUDIES, FBC, URINE MCS, GTT, HIV 1/2 AB
Tests Pending :TFT, RUBELLA IGG, CT NG PCR, GROUP ANTIBODY AND HOLD, HCG(QUANT)

FULL BLOOD COUNT
Collection Date: 08:40 03/12/2022
CUMULATIVE FULL BLOOD EXAMINATION

Date	23/03/22		03/12/22		
Time	09:36		08:40		
Lab No	69472786		71974230		
Hb	150		142	g/L	(115-160)
RCC	5.2		4.9	x10 ^12 /L	(3.6-5.2)
Hct	0.45		0.45		(0.33-0.46)
MCV	86		91	fL	(80-98)
MCH	29		29	pg	(27-35)
Plats	360		367	x10 ^9 /L	(150-450)
WCC	8.5		7.5	x10 ^9 /L	(4.0-11.0)
Neuts	4.4	59 %	4.4	x10 ^9 /L	(2.0-7.5)
Lymphs	2.4	25 %	1.9	x10 ^9 /L	(1.1-4.0)
Monos	0.5	6 %	0.5	x10 ^9 /L	(0.2-1.0)
Eos	1.19	9 %	0.68	x10 ^9 /L	(0.04-0.40)
Basos	0.00	1 %	0.08	x10 ^9 /L	(< 0.21)

71974230 Automated Comment:
As per ISLH guidelines - Film not reviewed. If a film review is truly indicated, contact the laboratory within 24 hours of collection. Otherwise investigate any highlighted abnormalities as clinically appropriate.

Eosinophilia, otherwise normal haematology parameters for pregnancy.

** FINAL REPORT - Please destroy previous report **

Tests Completed:HEP B / C, SYPHILIS AB, IRON STUDIES, FBC, GTT, HIV 1/2 AB
Tests Pending :TFT, RUBELLA IGG, CT NG PCR, URINE MCS, GROUP ANTIBODY AND HOLD
Tests Pending :HCG(QUANT)

GLUC. TOL. (2 OR 3H) PREG
Collection Date: 08:40 03/12/2022
GLUCOSE TOLERANCE TEST IN PREGNANCY
Oral glucose load: Adult 75 g

The Royal College of Pathologists of Australasia (Australian membership) has endorsed the IADPSG guidelines.

Time	serum Glucose	Diag. Threshold	urine Glucose	urine Ketone
Fasting	4.9 mmol/L	above 5.0	Not provided	Not provided
1.0 h	8.6 mmol/L	above 9.9		
2.0 h	7.0 mmol/L	above 8.4		

Interpretation:
Normal glucose tolerance.

Tests Completed:GTT
Tests Pending :HEP B / C, TFT, SYPHILIS AB, RUBELLA IGG, IRON STUDIES, CT NG PCR, FBC
Tests Pending :URINE MCS, GROUP ANTIBODY AND HOLD, HCG(QUANT), HIV 1/2 AB

HIV ANTIBODY TEST
Collection Date: 08:40 03/12/2022

HUMAN IMMUNODEFICIENCY VIRUS (HIV) TESTING

HIV antigen / HIV 1 HIV 2 antibody : Non reactive

In cases of possible HIV exposure, serology should be repeated in approximately 3 months.

The screening assay performed is an antigen/antibody combination test
which detects HIV 1, HIV 2 antibodies and HIV P24 antigen.

Tests Completed:IRON STUDIES, GTT, HIV 1/2 AB
Tests Pending :HEP B / C, TFT, SYPHILIS AB, RUBELLA IGG, CT NG PCR, FBC, URINE MCS
Tests Pending :GROUP ANTIBODY AND HOLD, HCG(QUANT)

IRON STUDIES
Collection Date: 08:40 03/12/2022

CUMULATIVE IRON STUDIES

Date	23/03/22	03/12/22		
Time	09:36	08:40		
Lab No	69472786	71974230		
Iron	19	16	umol/L	(10-33)
TIBC	64	61	umol/L	(45-70)
Saturation	30	26	%	(16-50)
Ferritin	46	53	ug/L	(25-290)

Tests Completed:IRON STUDIES, GTT
Tests Pending :HEP B / C, TFT, SYPHILIS AB, RUBELLA IGG, CT NG PCR, FBC, URINE MCS
Tests Pending :GROUP ANTIBODY AND HOLD, HCG(QUANT), HIV 1/2 AB

SYPHILIS SEROLOGY
Collection Date: 08:40 03/12/2022

SYPHILIS SEROLOGY

Treponemal antibody screen : Negative

Antibodies to Treponema pallidum NOT detected.

This result may indicate either no exposure to Treponema pallidum or very early primary syphilis.

If infection is suspected, please repeat in 14 days.

Tests Completed:SYPHILIS AB, IRON STUDIES, GTT, HIV 1/2 AB
Tests Pending :HEP B / C, TFT, RUBELLA IGG, CT NG PCR, FBC, URINE MCS
Tests Pending :GROUP ANTIBODY AND HOLD, HCG(QUANT)

HEPATITIS A B C
Collection Date: 08:40 03/12/2022
HEPATITIS SEROLOGY

Hepatitis B surface antigen (HBsAg) : Negative

Hepatitis C IgG antibody (HCVIgG): Negative

If contact with hepatitis is suspected, please repeat serology as indicated.
INCUBATION PERIOD (HEPATITIS A): 2-8 weeks
(HEPATITIS B): 1-6 months
(HEPATITIS C): 1-6 months

Tests Completed:HEP B / C, SYPHILIS AB, IRON STUDIES, GTT, HIV 1/2 AB
Tests Pending :TFT, RUBELLA IGG, CT NG PCR, FBC, URINE MCS, GROUP ANTIBODY AND HOLD
Tests Pending :HCG(QUANT)

Queensland Government

Townsville Hospital and Health Service

Medical Imaging Department
& Maternal Fetal Medicine Services

100 Angus Smith Drive
Townsville QLD 4814
T: 07 4433 1500

Apple Tree Medical Cairns
Michael BreitKreutz

MRN : 1039544-TTH
Date 18/04/2023

Thank you for your ongoing care with Jennifer (social worker at Innisfail hospital), a lovely 34 year old woman who recently delivered in our service on the 6th of April, 2023.

Jennifer presented to Cairns base hospital with unstable hypertension and bloods concerning of evolving HELLP. Jennifer and Zac did not want to fly and instead of RDFS retrieval flight, the parents insisted on driving.

The couple arrived on the 4th of April and was seen by Dr Iyer and was clinically stable. MET call at 1930 for hypertension 175/105 - requiring 5mg hydralazine, MgSo4 commenced. Repeat bloods showed Plts improved at 156, Hb 108, Creatinine = 69 and LFTs show ALT = 126 and AST = 34 (improved), normal bilirubin, LDH not ordered, normal coags. 2nd dose of steroids given and Jennifer had a consultation with the NICU team.

On a bedside ultrasound, there were concerns about reversed EDF in the UmA. I came in to ultrasound the baby and found a severely growth restricted baby (469 grams < 1st centile = 21+6 weeks), with minimal growth in the AC/HC/long bones since morphology scan. There was oligohydramnios and pre-terminal Doppler studies.

At the time of the ultrasound, I had a long discussion with Jennifer and Zac and the couple were keen not to deliver for fetal reasons, understanding the risk of stillbirth in the next 12 - 24 hours.
The next day, the baby was bradycardiac and had ongoing pre-termination Doppler studies. Further discussions with NICU and MFM, the couple requested termination of pregnancy. The couple then had a medical IOL and their little boy, Oliver, was born still, at 12:22 on the 6th of April. The couple were keen to proceed with a full autopsy.

The couple remained in hospital for 3 days for blood pressure management and was discharged on labetalol 400mg QID and methyldopa 500mg TDS.

We have discussed what a next pregnancy would like including
1) Preconception lifestyle changes
2) High dose folate
3) Early referral to our service after a dating scan confirming an IUP
4) We will arrange a 12 week ultrasound
5) To consider aspirin from 10 weeks onwards (150mg aspirin soluble at night)
6) High risk model of care

Yours Sincerely,

Dr Cecelia O'Brien
Provider No: 2885298L.

Dr Cecelia O'Brien
MFM Staff Specialist

Sonographer

CC: Dr. Samantha Sherman, Consultant Obstetrician, Cairns; Dr. Dianne Payton, Perinatal Pathologist, Herston