Wellington Regional Genetics Laboratory

Level 6, WSB, Wellington Hospital
Phone: 04-918-5352
Fax: 04-385-5822
Email: wrgl@ccdhb.org.nz
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IANZ

PRENATAL DIAGNOSIS REQUEST FORM

PATIENT & REFERRER (to be completed by the obstetrician)

Patient Name: ……

Patient DOB: …… NHI No: ……..

Address: ……

Consultant's location: ….....…….……

Obstetrician: ……..…

Email: ……Ph: ……

Lead Maternity Provider: …….

Email: ……Ph: ……

Additional copies of report to: ……

FAMILY / PREGNANCY HISTORY (to be completed by the obstetrician)

No. of previous pregnancies: …… No. of children: …….. Miscarriages: …….....

Amnio/CVS in previous pregnancy? ☐ Yes ☐ No Under what name/where:.....

INDICATION FOR TEST EDD: ……..…… Gestation: ..…….....

Screening risk 1: …… for …….. ☐ MSS1 ☐ Combined (MSS1+NT) ☐ MSS2 ☐ Integrated (MSS1/2+NT)

☐ NT alone NT measurement mm: ……

☐ High risk NIPT result. High risk for ……. copy of NIPT report attached ☐

☐ Maternal age / Anxiety

☐ Abnormalities on scan: specify……..……..……

……..…

☐ Family member with genetic condition or known genetic abnormality i.e. parent, partner, sibling, children etc :

Name: ..... DOB: ……..…. Condition: ….....

☐ Family translocation: specify: ……..…….

☐ Other ……..……..

TEST

☐ Microarray + QF-PCR Array consent form (PTO) & 4ml parental bloods req'd (EDTA & Li Hep)

☐ Karyotype + QF-PCR

☐ Molecular Test ** + QF-PCR Gene: …… ** Molecular consent form required

SAMPLE

☐ CVS Weight (est): ……. Sample condition ……..……..

☐ Amniocentesis Volume: .....……Clear ☐ Bloodstained (inc 4ml EDTA blood) ☐

For lab use only:

Obstetrician's signature: ……..…

Sample Date: ……..

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MICROARRAY CONSENT AND INFORMATION

Genetic File No. ……

For lab use only:

Consent is given for:

1. **Chromosomal Microarray (CMA)**

   Sample Type: CVS/Amniocentesis

   Laboratory: Wellington Regional Genetics Laboratory

2. **Sample Storage**

   Sample Type: DNA and Fixed Cell Storage

   Laboratory: Wellington Regional Genetics Laboratory

3. **Participation in Microarray Database Repository**

   Study: DECIPHER

   Location: Cambridge UK https://www.deciphergenomics.org

The information from this test may be used for other family / whanau (members) to benefit from genetic testing, if you do not wish to do this, please state below:

NOTE: Named test results will not be divulged without further consent.

I understand that this sample and/or any results will not be released to any other third party without my consent (or unless legally required).

I am aware that genetic testing may have insurance implications.

In some circumstances, testing may reveal information about biological relationships.

On rare occasions, genetic testing may reveal findings we were not anticipating that are not related to the condition discussed. This will be discussed with you should this occur.

This sample may be used if additional testing is indicated for this condition in the future.

DNA may be returned or destroyed (contact WRGL to arrange).

I have read and understood the information given to me and have had the opportunity to ask questions. I understand that I may withdraw or modify this consent at any stage, and that such withdrawal will not affect my further health care.

Signed:		Date:
	Patient / parent or guardian / or next of kin	
	Medical Professional	

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Information for Parents about Microarray Analysis

Your doctor has requested a chromosomal microarray analysis for your pregnancy. This test is used to help identify a genetic cause for physical, behavioural or intellectual differences.

Microarray analysis is a very detailed test looking for small changes in the amount of genetic information, which may be missed by a standard chromosome test, using a microscope.

The microarray result will be one of the following:

- **No clinically significant change i.e. a normal result**
  No clinically relevant chromosome change has been detected. CNVs that are classified as susceptibility loci are not routinely reported in the prenatal setting unless their penetrance is estimated to be >20%, or they are associated with the anomaly (or anomalies) detected on ultrasound scan. Re-evaluation can be requested if further clinical information is available. A full list of these CNVs is available to the clinician through contacting the laboratory.
  This result does not exclude all genetic conditions.

- **Pathogenic change i.e. abnormal result**
  A chromosome change has been detected which may cause physical, behavioural or learning differences.

  Very rarely a chromosomal abnormality is found that has important health implications but is not related to the reason for referral e.g. the result may be relevant to genetic conditions that have an onset in later life or those that are associated with an increased risk of cancer (*Unsolicited finding of clinical significance*).

- **Change of uncertain/unknown significance**
  A chromosome change has been detected, but there is limited information available about the effects of the chromosome change and therefore it is difficult to know whether the change is the cause the findings associated with your pregnancy.

  A change may be associated with variability in the problems it can cause i.e. different people may be affected more or less severely, and it may not be possible to provide the exact risk of a change causing problems or how significant the problems will be.

In some cases samples from each parent may also be analysed to help to assess the importance of the change within the family. Interpretation of a finding may be more difficult if samples from both parents are not available.

Some additional possible outcomes of testing are:

- A change may be detected, which may subsequently be found in either parent
- The test may reveal information about biological relationships
- The test may show that the child (and / or parents) is a carrier of a recessive genetic disorder. In this situation, genetic counselling will be required to ensure the family understand the implications of the results

Due to the rarity of some of the chromosome differences the laboratory participates in an international group, which provide a resource about these various chromosome differences and the clinical features of individuals with those differences. The databases collect information about the microarray result, and the physical, intellectual or behavioural characteristics of the person with the abnormality. This information is submitted without the identifying information about the person. Participation in these databases is important for the assessment of the individual changes, and also for the future analysis of the test.