

**FERNIE, Larissa Ellen (Mrs)**

456 James Terrace  
Sunrise Bay QLD 4573

Sex **Female**

DOB **28-May-88, aged 32y**

MRecord **719909917**

Located at Sunrise Hospital  
Fertility Clinic

Request ID **1978881774**

on **20-Sep-19**

by **Dr Trish FAMILYDR**

733 Little Archie Street  
Brisbane QLD 4001  
Provider 951577QT

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**Chromosome studies – Final Report**

**Clinical history** – Primary infertility

**Report issued**

**26-Sep-19 10:00**

Collection date Collection time Request ID <b>Specimen</b>	<b>Latest results</b>		
	<b>25-Sep-19</b>		
	<b>08:20</b>		
	<b>1978881874</b>		
	<b>Blood</b>	Reference	Units
Cytogenetic karyotyping	46,X,del(X)(q11.2)		

**Results**

An abnormal female result showing a deletion of the long arm of the X-chromosome at band q11.2.

This aberration was observed in all cells analysed.

GTG banded analysis was performed on 5 cells analysed and 10 cells counted at a resolution of 550 bands.

**Interpretation**

Deletions of the long arm of the X-chromosome are associated with a variable phenotype in females. Common findings include short stature, gonadal dysgenesis and premature ovarian failure. It is likely that this aberration is the cause of the primary infertility observed in this patient. Genetic counselling is recommended.

Dr Kondos Pathologist

+61 7 3662 9871

Copy to 1 – Genetic Counselling Clinic coordinator

Copy to 2 - Dr Sarsgaard Genetic Counsellor, 81 Estelle Parade, New Farm

Copy to 3 - MyHealthRecord, IHI 8003 6063 6173 7731

For consumer information on pathology results visit Lab Tests Online Australia <https://www.labtestsonline.org.au/>

**PITUS Pathology Sunshine Coast Laboratory**

8-12 Birdsong Drive MARCOOLA QLD 4573

**Medical Director:** Dr Gina Director Provider 355812C

Results enquiries: 1300 112 221 or [www.pituspath.com.au](http://www.pituspath.com.au)

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