Fact Sheet

Factor V Leiden and Factor II (Prothrombin) Mutation Testing

The **Factor V Leiden mutation** is the most common cause of familial venous thrombosis with an incidence of about 3% in the general Caucasian population. Inheritance is in an autosomal dominant fashion.

Heterozygosity and Homozygosity for the Factor V Leiden mutation is associated with an estimated 8-fold and 100-fold increased risk of venous thrombosis, respectively.

The Factor V Leiden mutation is seen in 20-50% of thrombosis patients. Pregnancy and oral contraceptives can also increase the risk of venous thrombosis significantly.

The **Factor II (Prothrombin) mutation** is inherited independently of the Factor V Leiden mutation and in an autosomal dominant fashion. The incidence of the mutation is about 2% in the general Caucasian population.

Heterozygosity for the G20210A prothrombin mutation is associated with an approximate 3-fold increased risk of venous thrombosis. Heterozygosity for both Factor II and V mutations confers an increased risk of venous thrombosis by approximately 30-fold.

The Diagnostic Genomics Laboratory offers both Factor V Leiden and Factor II (Prothrombin) mutation testing daily during business hours in a single test (Cepheid GenXpert FII/FV Assay). Results are typically available within 48 hours.

The required specimen is 4-10mL of EDTA whole blood.

For further information please contact:

Dr Dipti Talaulikar	*Specialist Haematologist*	*51242487*
Fiona Webb	*Chief Scientist*	*51243449*

Phone 02 5124 2932 Fax 02 5124 2815
actpathology@act.gov.au
www.health.act.gov.au/services/act-pathology
Gilmore Crescent Garran ACT 2605 PO Box 11 Woden ACT 2606

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