

## Hereditary diffuse gastric cancer – OMIM 137215

### Background

Hereditary diffuse gastric cancer is caused by mutations in the *CDH1* (OMIM 192090) and *CTNNA1* (OMIM 116805) genes. The gene panel that includes these two genes also encompasses lobular breast cancer as a phenotype, in addition to diffuse gastric cancer. Pathogenic variants within the *CDH1* gene are associated with a lifetime risk of up to 56-70% and ~40% for developing diffuse gastric cancer and lobular breast cancer, respectively. Mutations in *CTNNA1* are also known to increase susceptibility to gastric cancer, however due to the penetrance of the gene, the level of increased risk is still unknown.

### Molecular Analysis

Testing of the *CDH1* and *CTNNA1* genes can be requested by clinical genetics, gastroenterology and gastroenterology surgeons.

**Next Generation Sequencing (NGS):** Analysis of the *CDH1* and *CTNNA1* genes for sequence and copy number variants, sequenced on an Illumina sequencing platform.

**Family follow-up:** Testing for known familial pathogenic variants in *CDH1* or *CTNNA1*

### Turnaround Times (TAT)

Prices available on request, please contact the laboratory using details below.

Test	TAT (calendar days)
Next generation sequencing and dosage analysis ( <i>CDH1</i> and <i>CTNNA1</i> )	42
Testing for known familial variant in <i>CDH1</i> and <i>CTNNA1</i>	28

#### Contact Details

All Wales Genomics Laboratory,  
Wales Genomic Health Centre,  
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Whitchurch,  
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[lab.genetics@wales.nhs.uk](mailto:lab.genetics@wales.nhs.uk)  
<http://www.wales.nhs.uk/AWMGS/>

#### Sample Requirements

Blood: 5ml in EDTA (1ml neonates/infants).  
Please contact lab prior to sending a prenatal sample.

Please label samples with three identifiers and date of collection  
**All samples must be accompanied by request form**  
Consent for testing & DNA storage is assumed when request for test received

#### Links

Orphanet

<http://www.orpha.net/>

OMIM

<http://www.omim.org/>

Genetic Test Registry

<http://www.ncbi.nlm.nih.gov/gtr/>