HNF1B genetics

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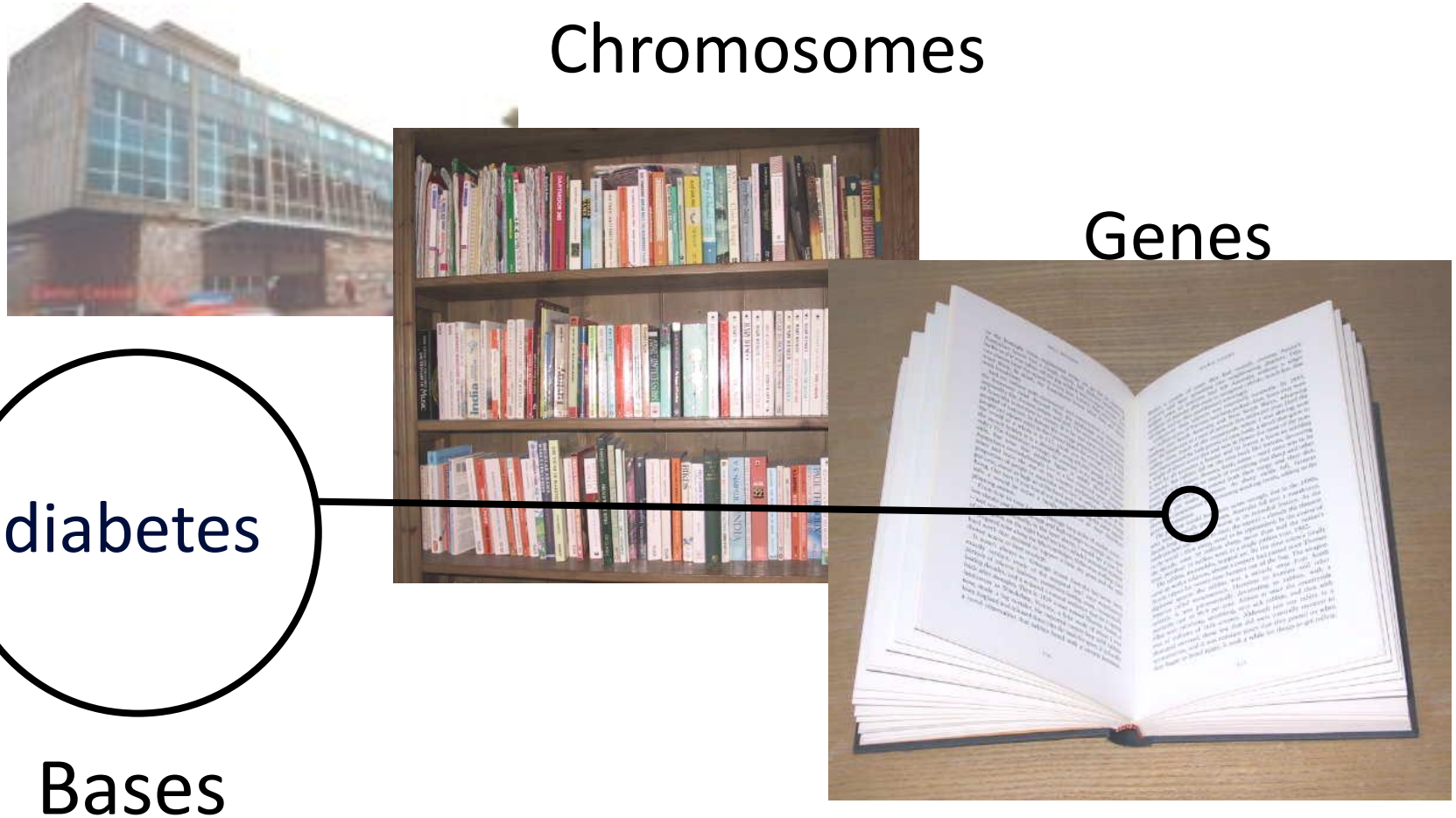
www.diabetesgenes.org

Human Genome

3,000,0000,000 bases
Each bases A, C, G or T

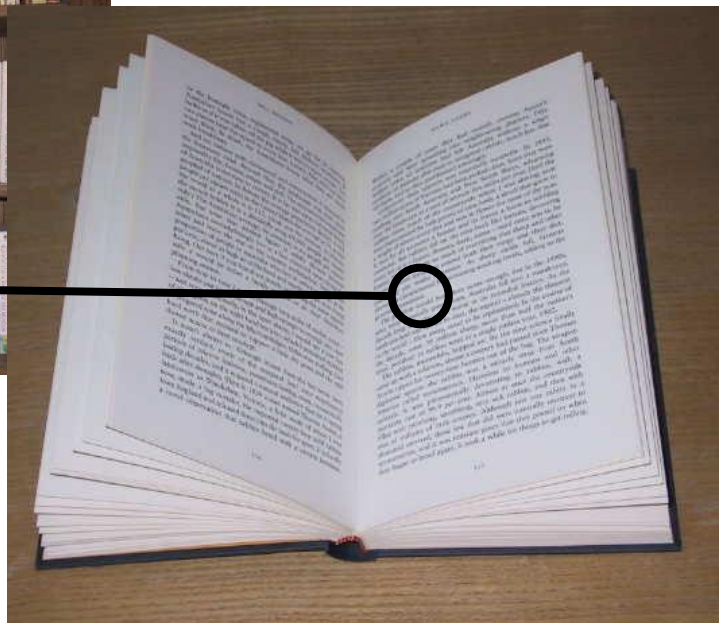
As much information in a single cell of the human body as there are letters in a library

The human genome: the human library



HNF1B disease can result from a variant or mutation –a single misspelt word in the library

1 change in 3,000,000,000 bases



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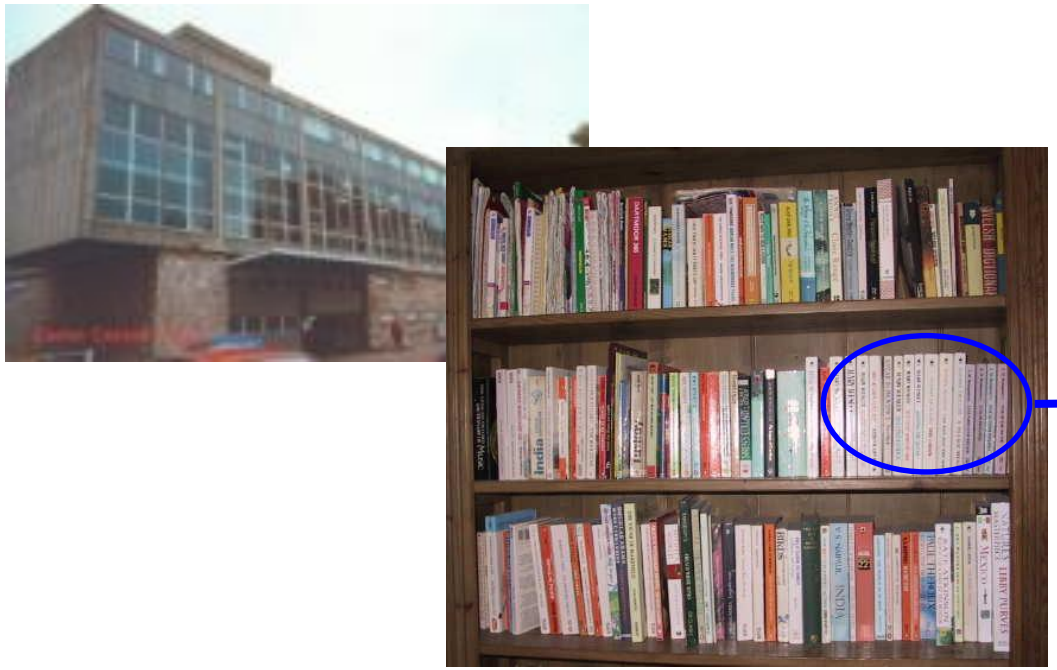
OR HNF1B disease can result from 17q12 deletion where 1 copy of 15 books removed from one bit of one bookshelf- this includes the HNF1B book



15 books have 1 copy removed

For every gene (book) you have 2 copies and only 1 of the 2 copies of HNF1B is misspelt or absent

The deletion or faulty copy of HNF1B may be inherited from an affected parent

OR in about half of cases it may be a new problem (de novo) when neither parent is affected

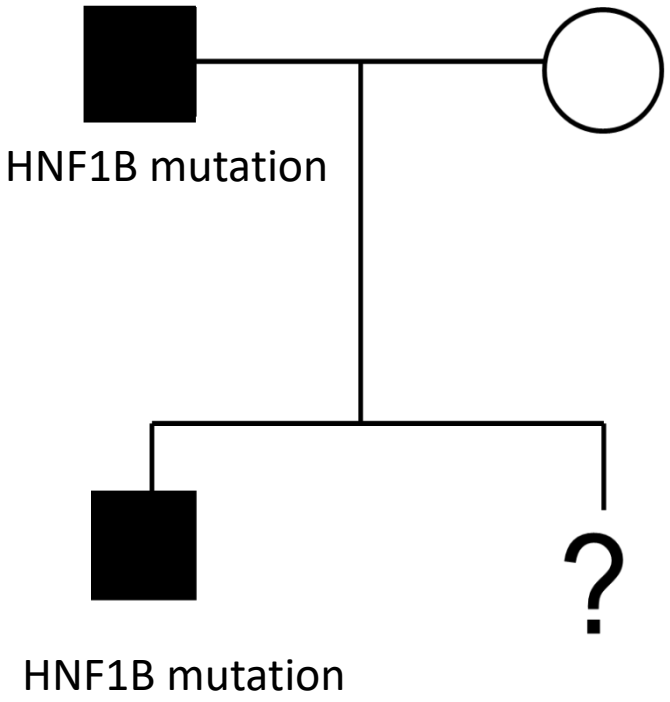
For every gene (book) you have 2 copies and only 1 of the 2 copies of HNF1B is misspelt or absent

When a person with HNF1B has a child they pass on one copy of each gene to their children

1 in 2 chance that a child will get the HNF1B disease causing version of the gene

Inheritance of HNF1B disease

- Will my next child inherit an HNF1B mutation?



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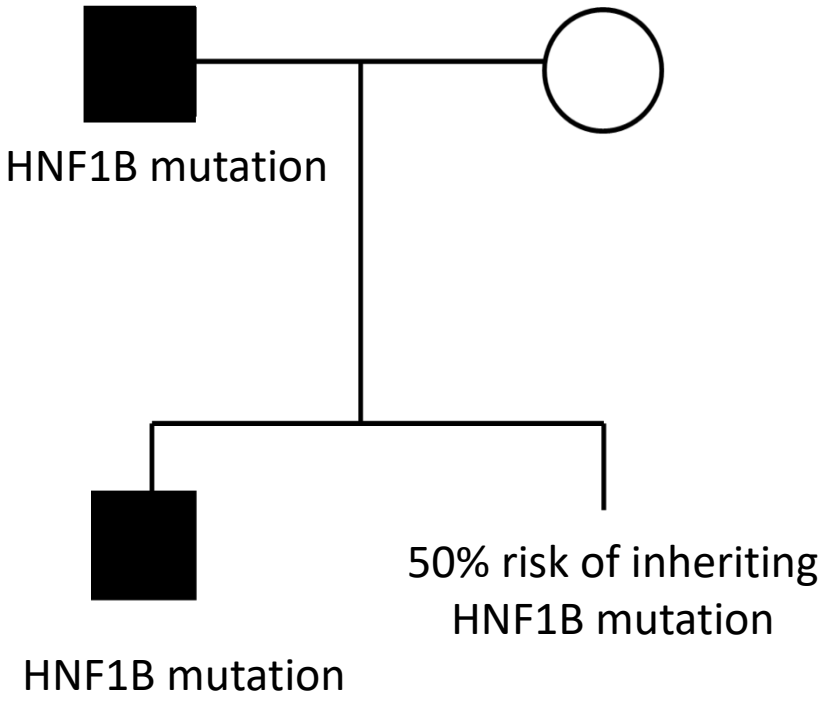

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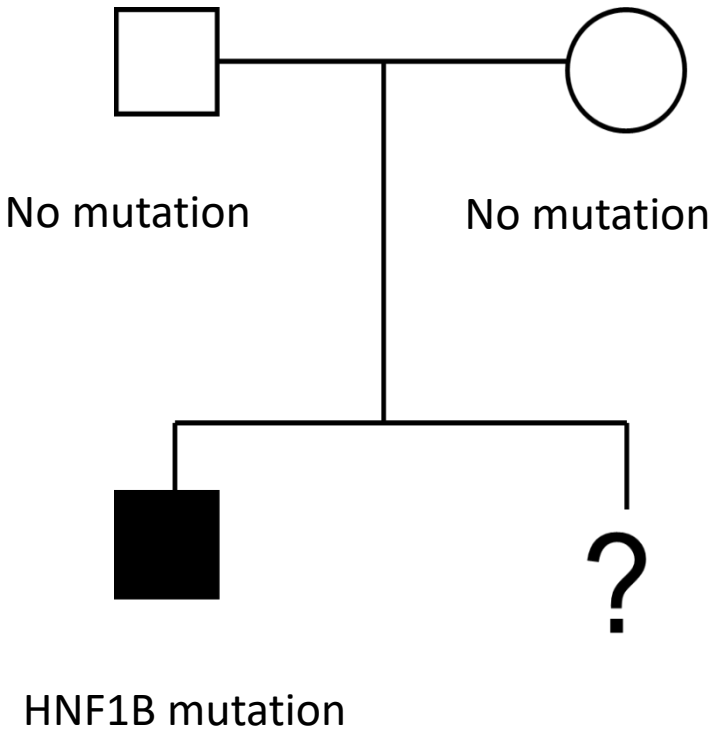

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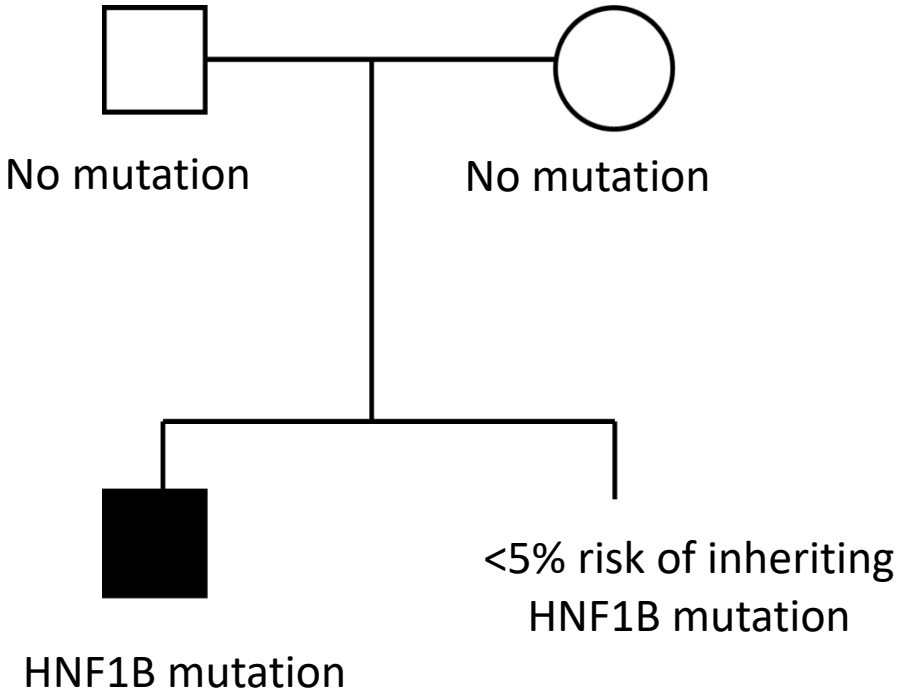
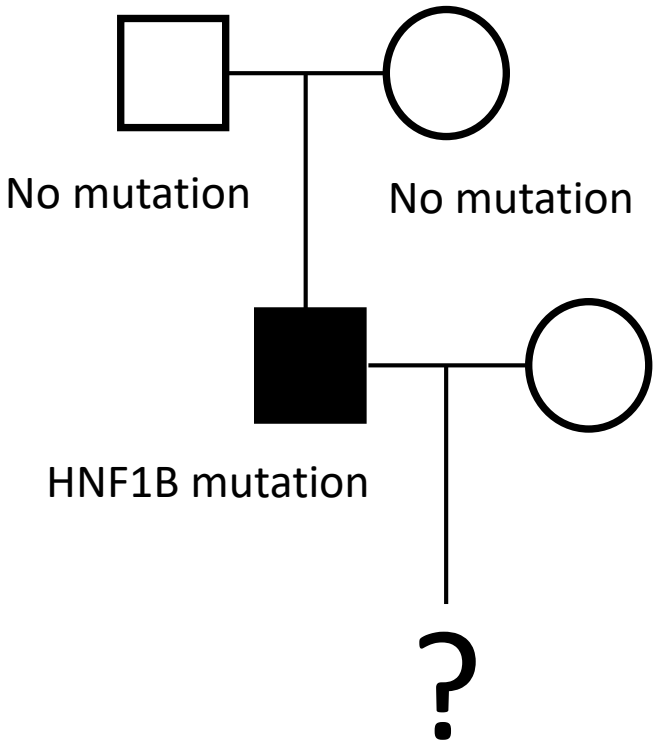


Inheritance of HNF1B mutations

- Will my grandchild inherit an HNF1B mutation?



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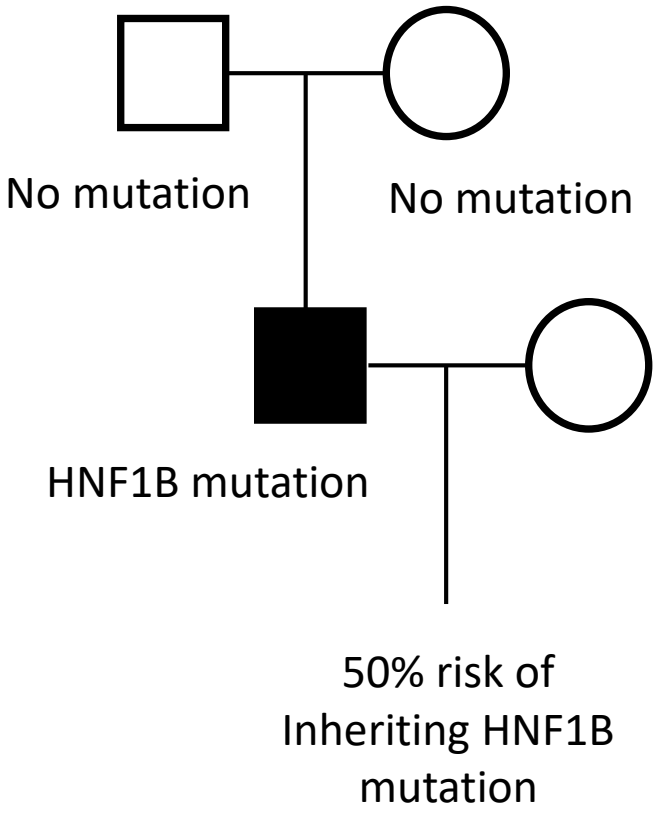
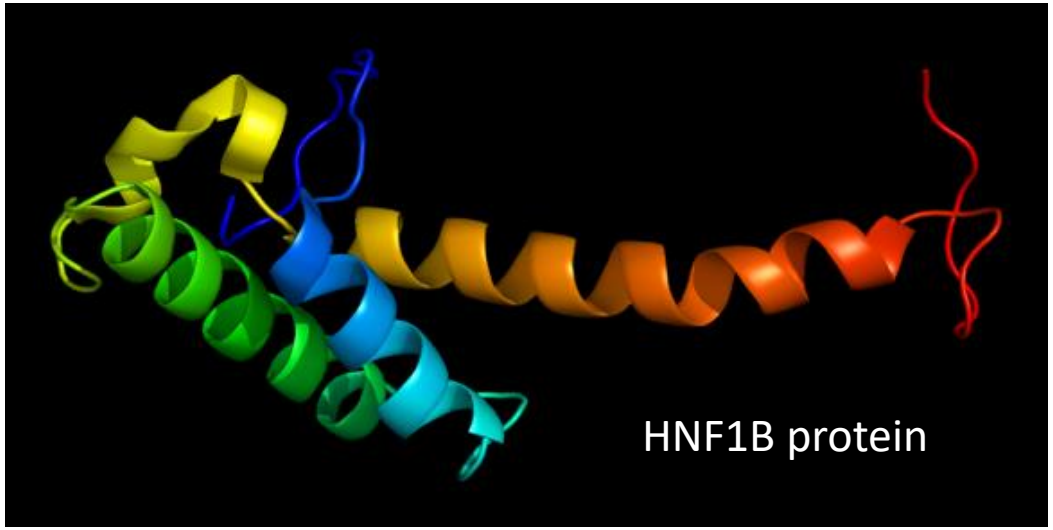


HNF1B (Hepatocyte Nuclear Factor 1 Beta)

- What is it?
- What does it do?

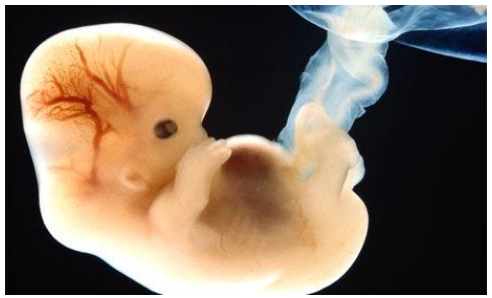


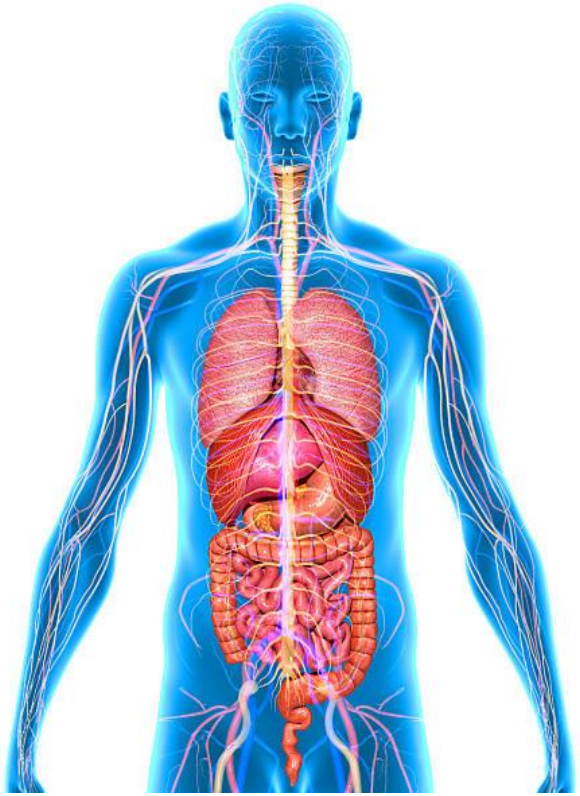
HNF1B: A transcription factor

- Transcription factors switch genes on or off (genetic “traffic lights”)

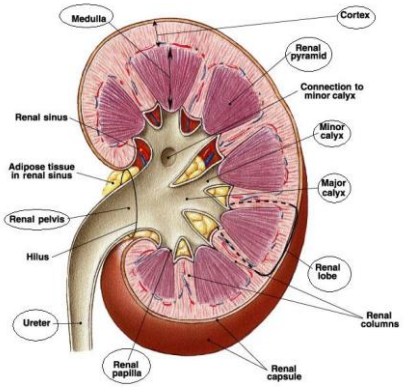
HNF1B: A transcription factor

- Transcription factors are key for development of the human body and all its parts

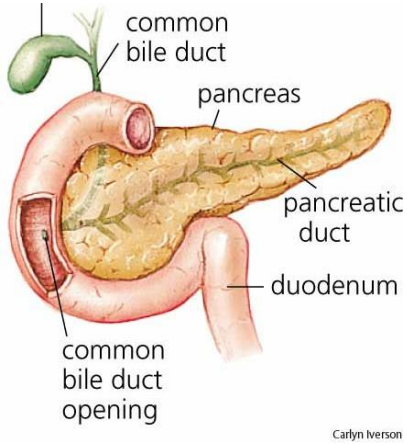




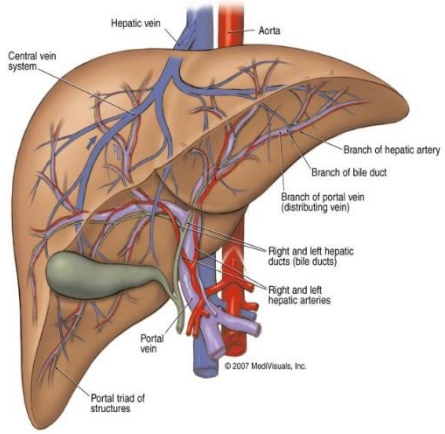
HNF1B is important in the making many parts of the body



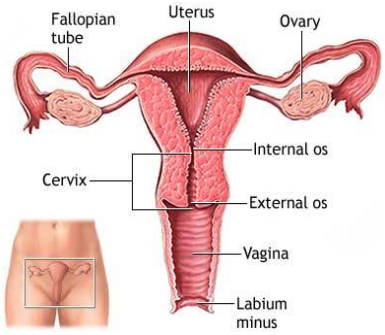
Kidney



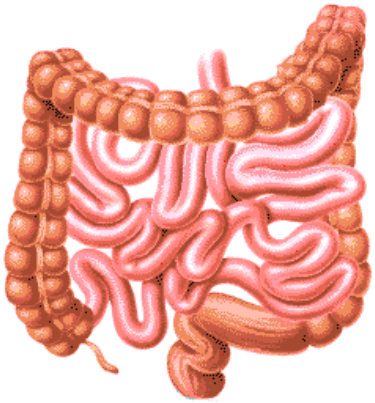
Pancreas



Liver



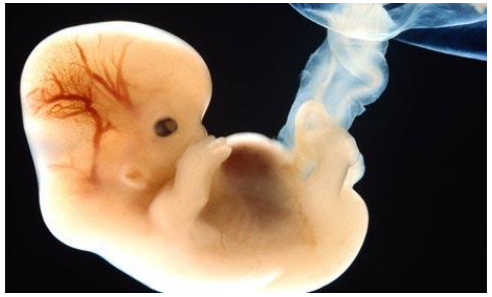
Female genital tract

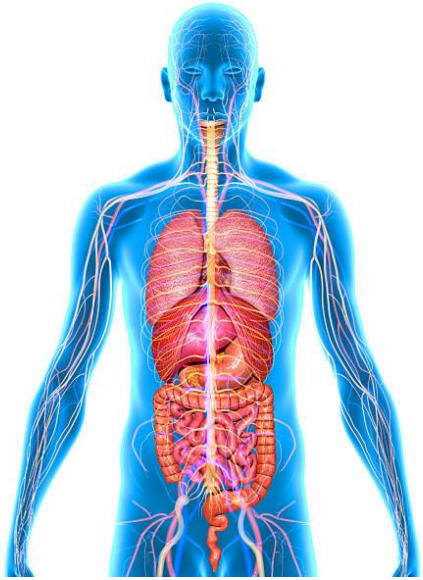


Gut

The effect of HNF1B on early development is variable

- This explain why even within a family with the same genetic variant the organs involved and the severity of their involvement can be very different





The Genetics of HNF1B disease

Either a spelling mistake in the gene
(mutation or variant)
OR a loss of one copy of the whole gene (deletion)

50% of offspring (on average) will be affected

HNF1B is a gene that controls other genes
especially genes involved in early development
especially in kidney & pancreas