

# HNF1B genetics

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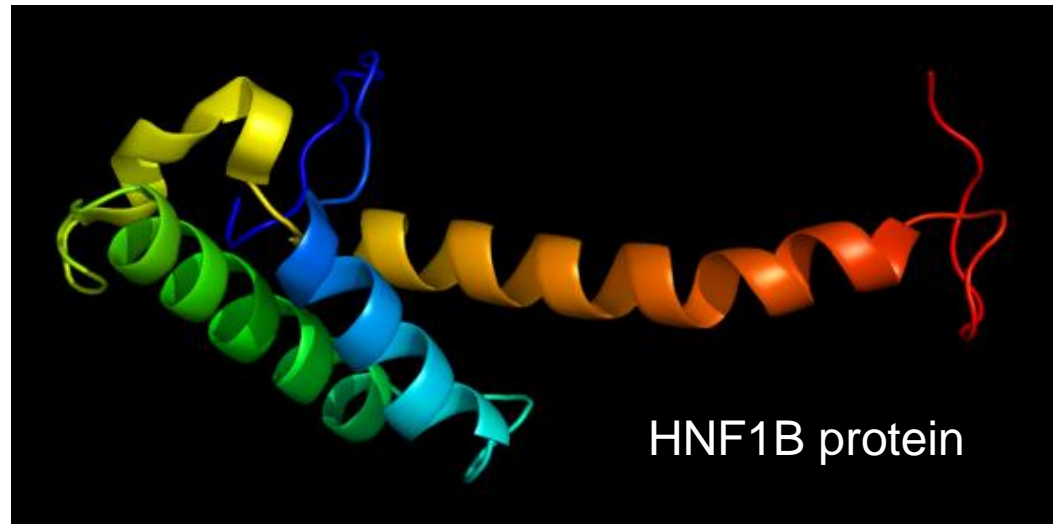
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[www.diabetesgenes.org](http://www.diabetesgenes.org)

# 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”)

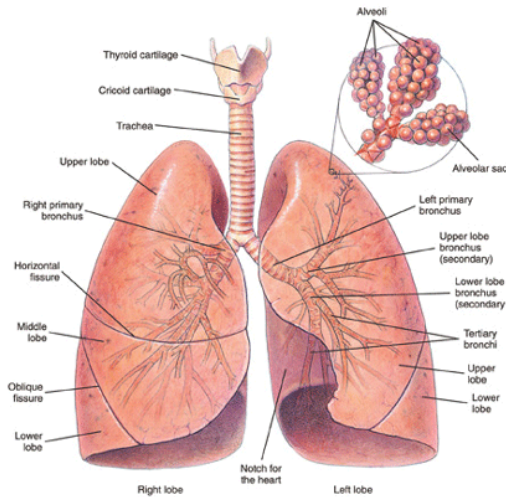


# HNF1B: A transcription factor

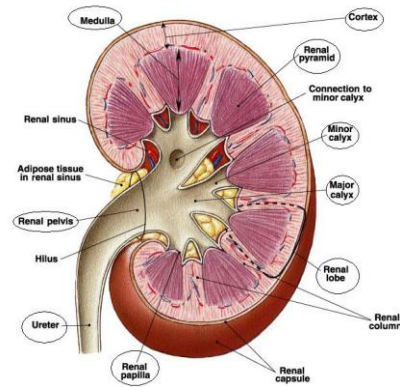
- Transcription factors are key for development of the human body



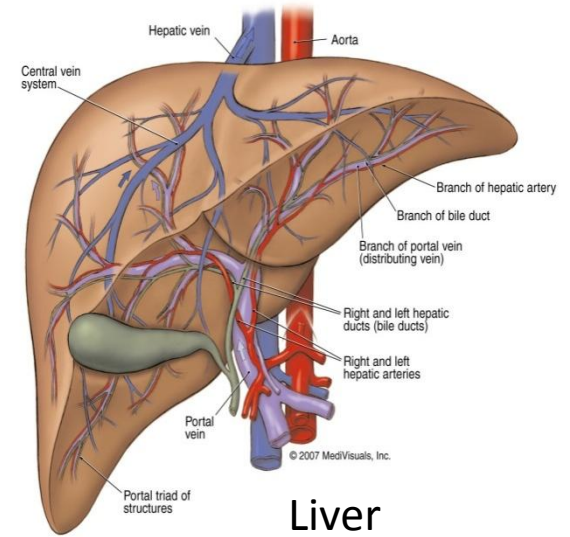
# HNF1B is found in many parts of the body



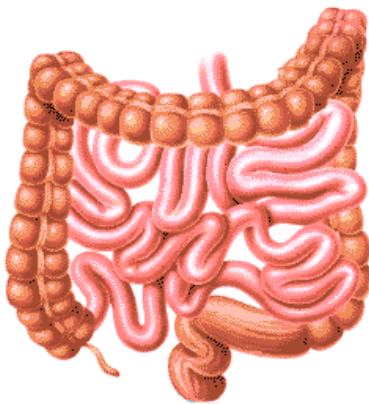
Lung



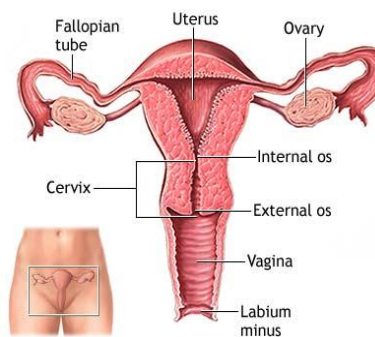
Kidney



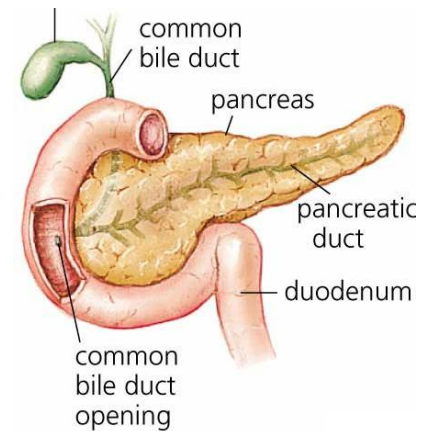
Liver



Gut



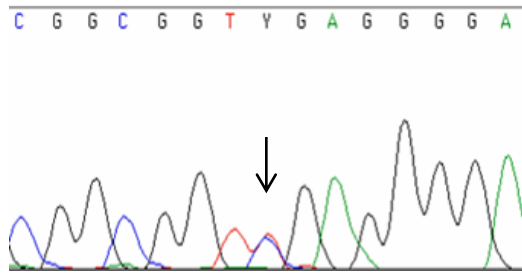
Female genital tract



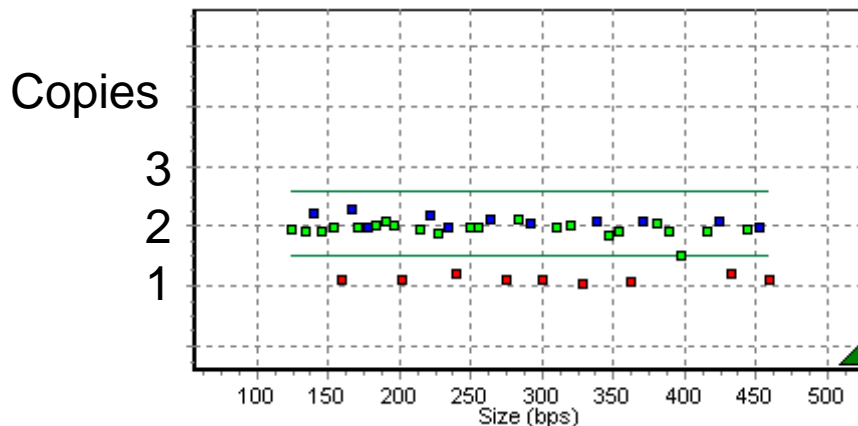
Pancreas

# HNF1B mutations causing disease

- Genetic “spelling mistakes”



- Gene is missing

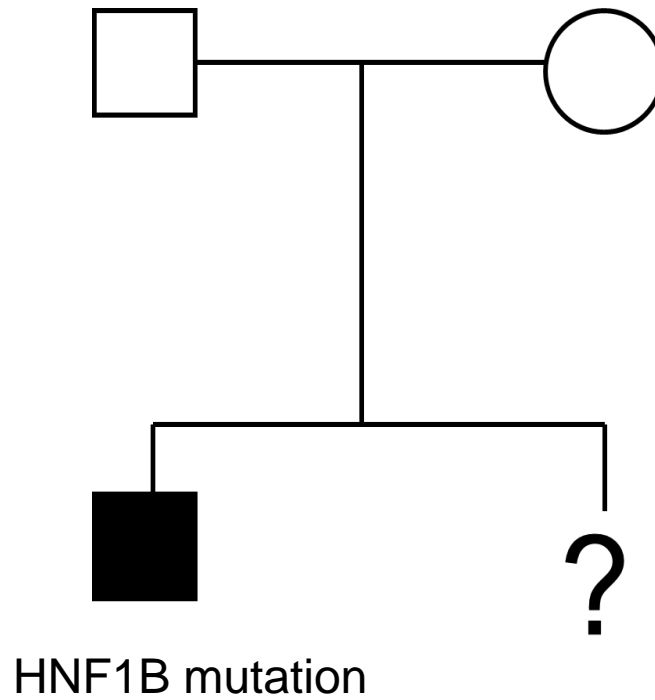


■ HNF1A  
■ HNF4A  
■ HNF1B



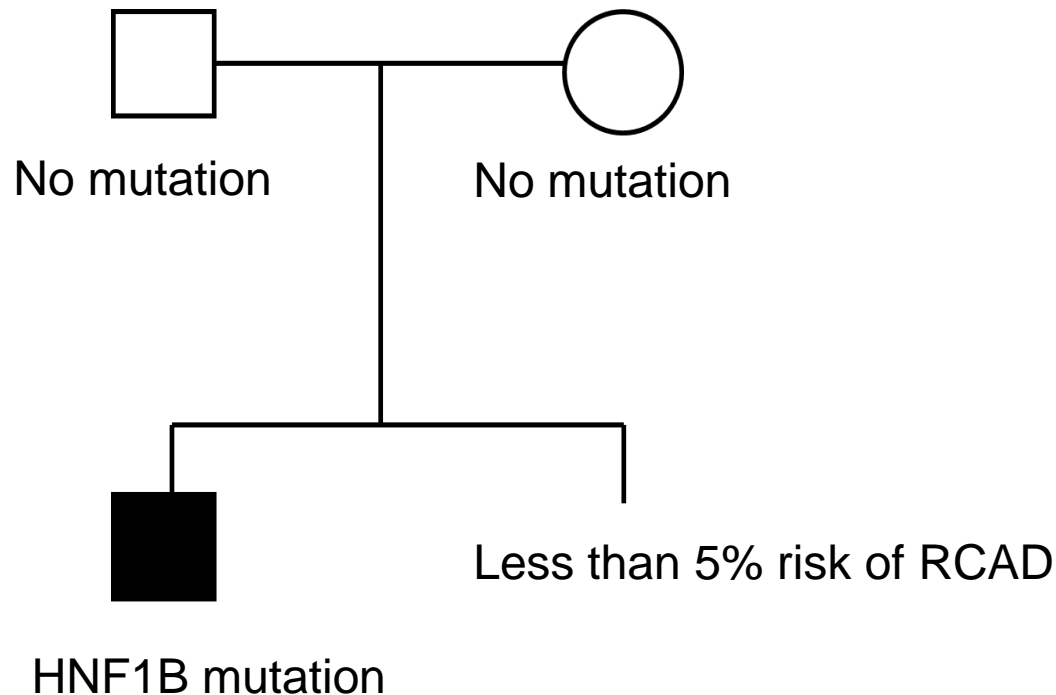
# Inheritance of HNF1B mutations

- Will my next child have RCAD?



# Inheritance of HNF1B mutations

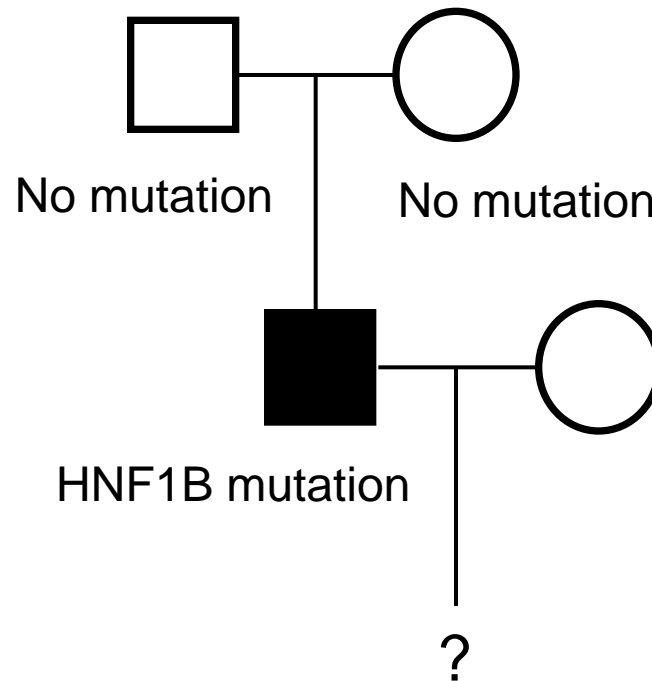
- Will my next child have RCAD?





# Inheritance of HNF1B mutations

- Will my grandchild have RCAD?



# HNF1B mutations causing disease

- Will my grandchild have RCAD?

