

What does dominant with incomplete penetrance mean?

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DEFINITION: DOMINANT WITH INCOMPLETE PENETRANCE:

Mutations that do not fit a simple pattern of Mendelian inheritance, and show variable degrees of clinical symptoms (variable expressivity of the gene) are said to be inherited with incomplete penetrance. Simply put, this means that individuals with one or two copies (in the case of dominant mutations) can appear to be normal, but can pass on a trait with varying degrees of clinical manifestations to their offspring. In the case of JRD this can be from normal to having a positive biopsy with no clinical signs of kidney dysfunction to severely affected. This is also consistent with the wide range of biopsy findings (% of fetal glomeruli) and individuals with a relatively high percentage of fetal glomeruli, that are still able to concentrate urine.