

Is the genetic diagnosis process similar for all rare diseases?

In this video, Prof Laurence Faivre explains how the diagnostic process is highly dependent on the disease and which are the main situations that can be met.

Watch and complete:

1- How many different genetic diseases are there?

2- Which examples are mentioned?

- the diagnosis is obvious and a simple test can confirm it:
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- the disease can be heterogeneous with many genes involved:
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3- Which would be the most suitable for an unlabelled malformative syndrome with an intellectual disability?