

3q29 Deletion Syndrome: Medical and Physical Characteristics



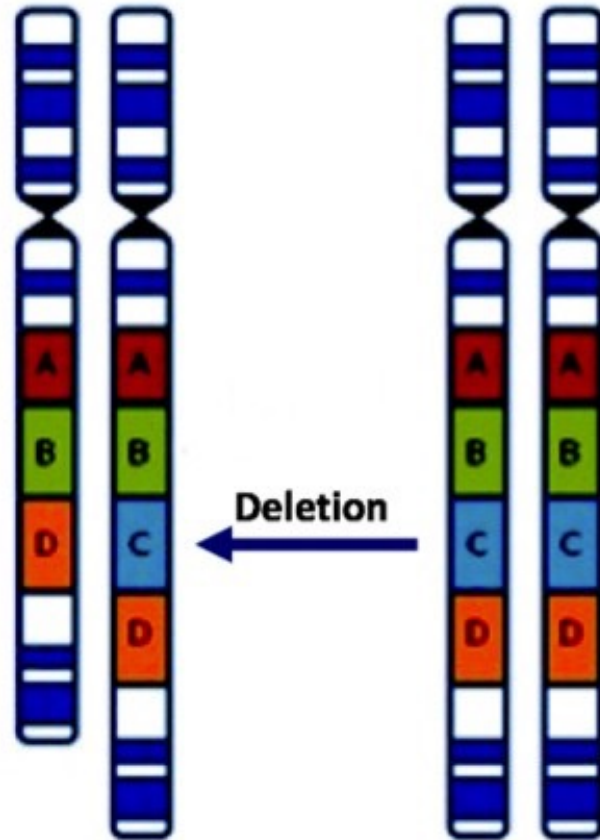
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What is a Copy Number Variant?



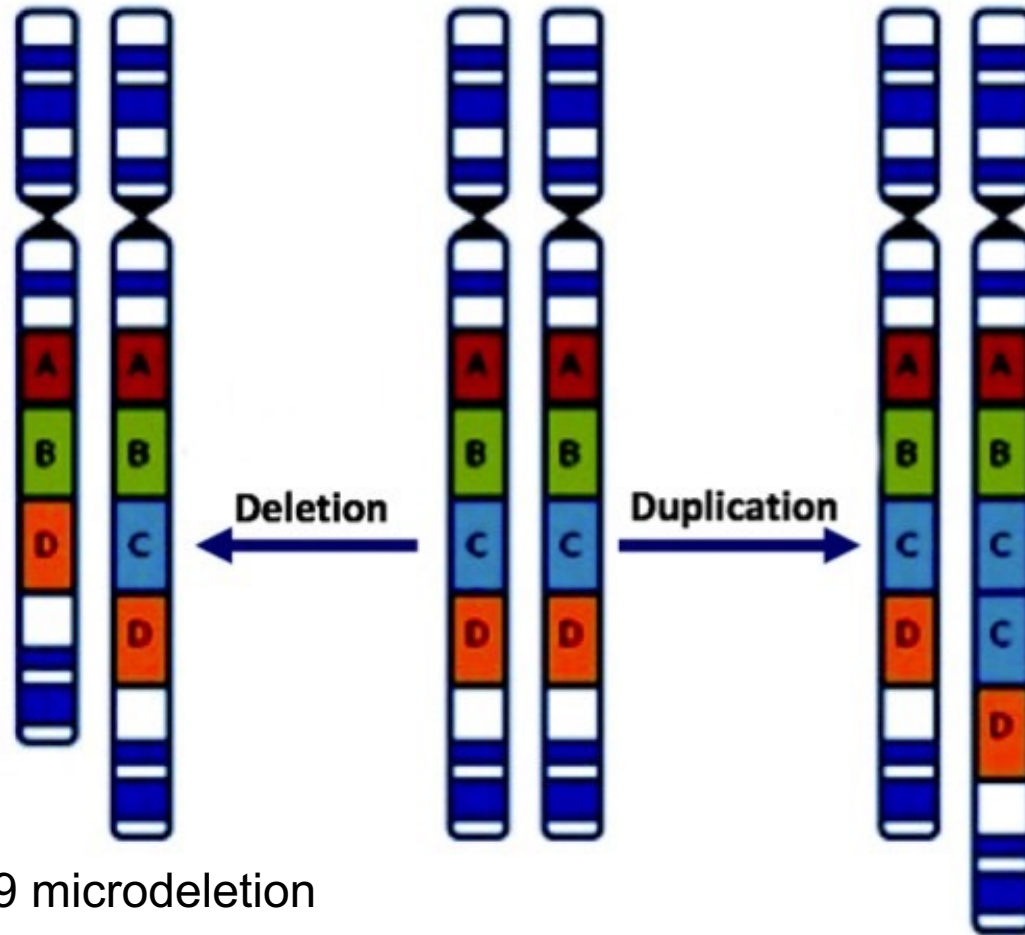
3q29 deletion = 3q29 microdeletion

What is a Copy Number Variant?



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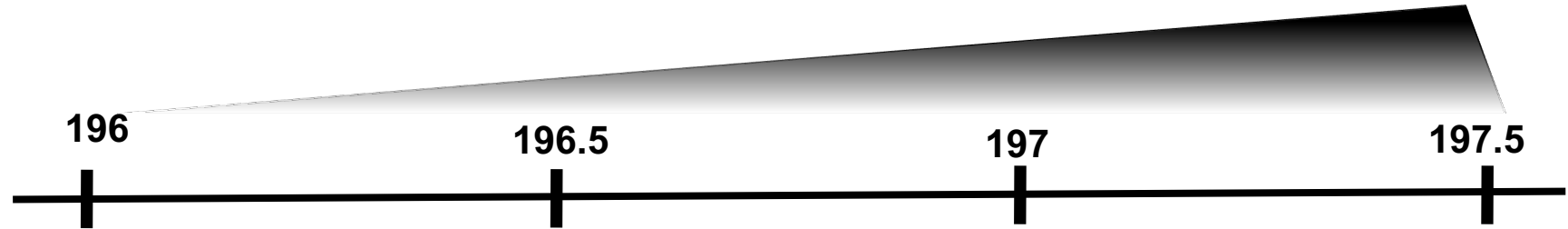


3q29 deletion = 3q29 microdeletion

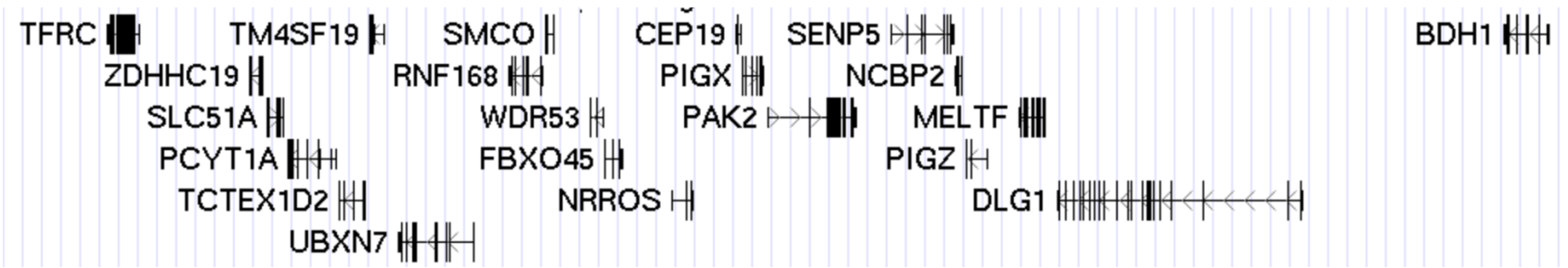
What is the 3q29 deletion?



Mb on chr3q:



21 Genes in the Interval:



Typical deletion:



Low-Copy Repeats:



00:05:53

Rare Syndrome: 1 in 30,000 in the general population

First description of the 3q29 deletion:



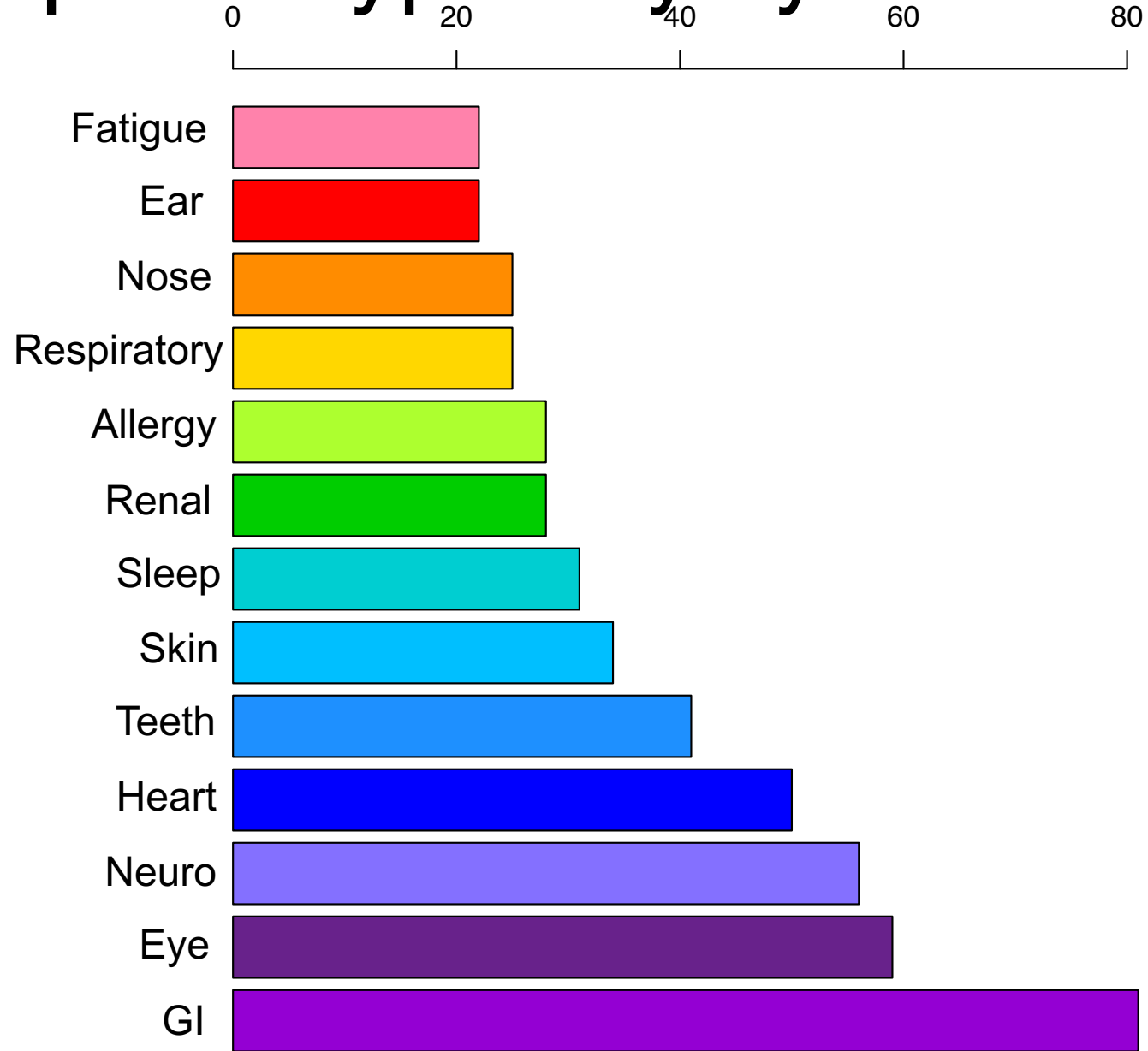
3q29 Microdeletion Syndrome: Clinical and Molecular Characterization of a New Syndrome

- Willatt et al, AJHG 2005 describes six patients
- All have mild to moderate ID and language delay
- “Autism was a feature of the behavior of two of the patients”

Expanding the clinical phenotype of the 3q29 microdeletion syndrome and characterization of the reciprocal microduplication

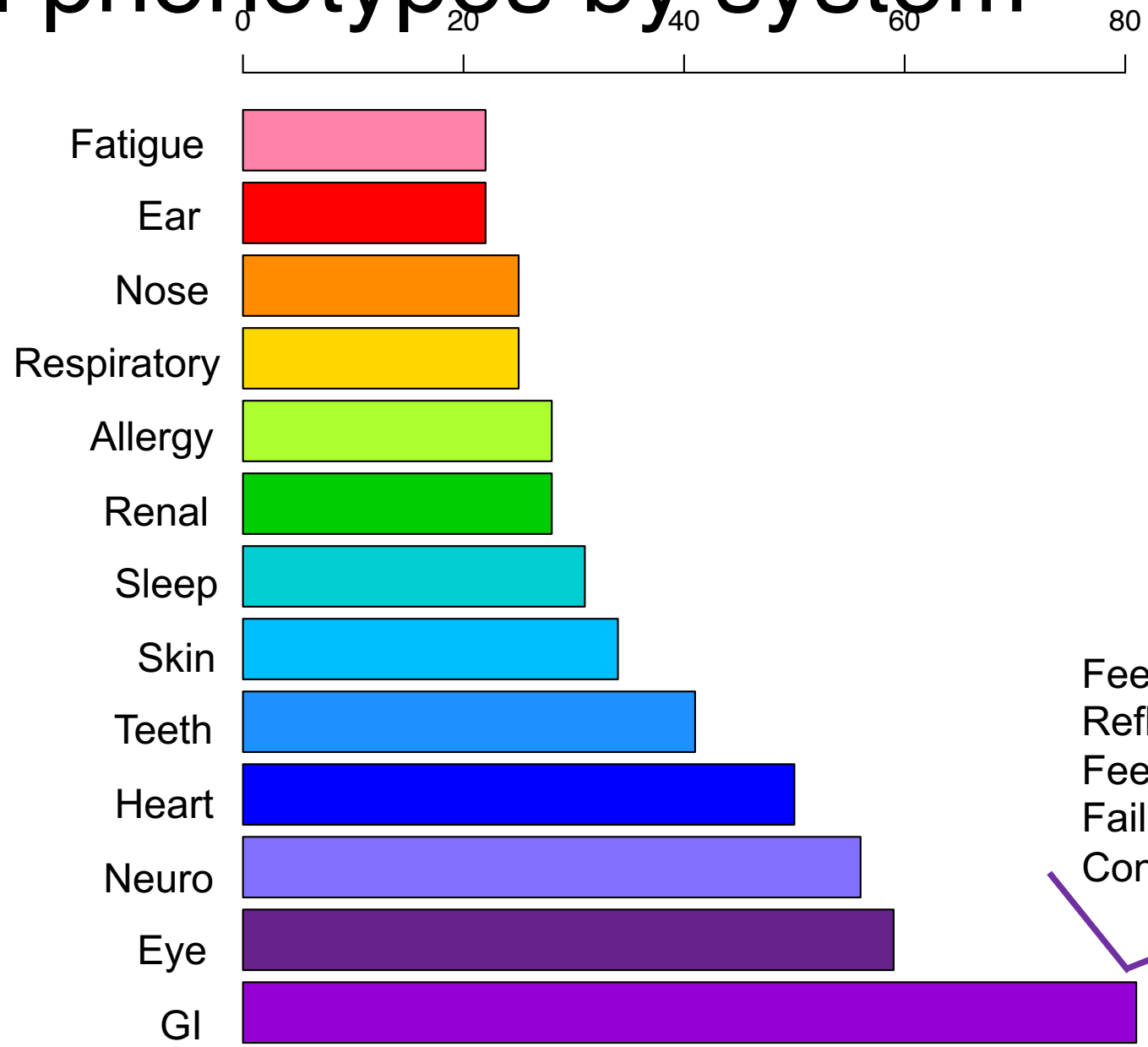
- Ballif et al 2008, Mol Cytogenetics: 14 patients
- Mild to moderate ID the only feature common to all patients
- Speech Delay (53%)
- Autism/autistic features (27%)
- Recurrent ear infections, heart defects, widely spaced teeth (13%)

Medical phenotypes by system



00:08:57

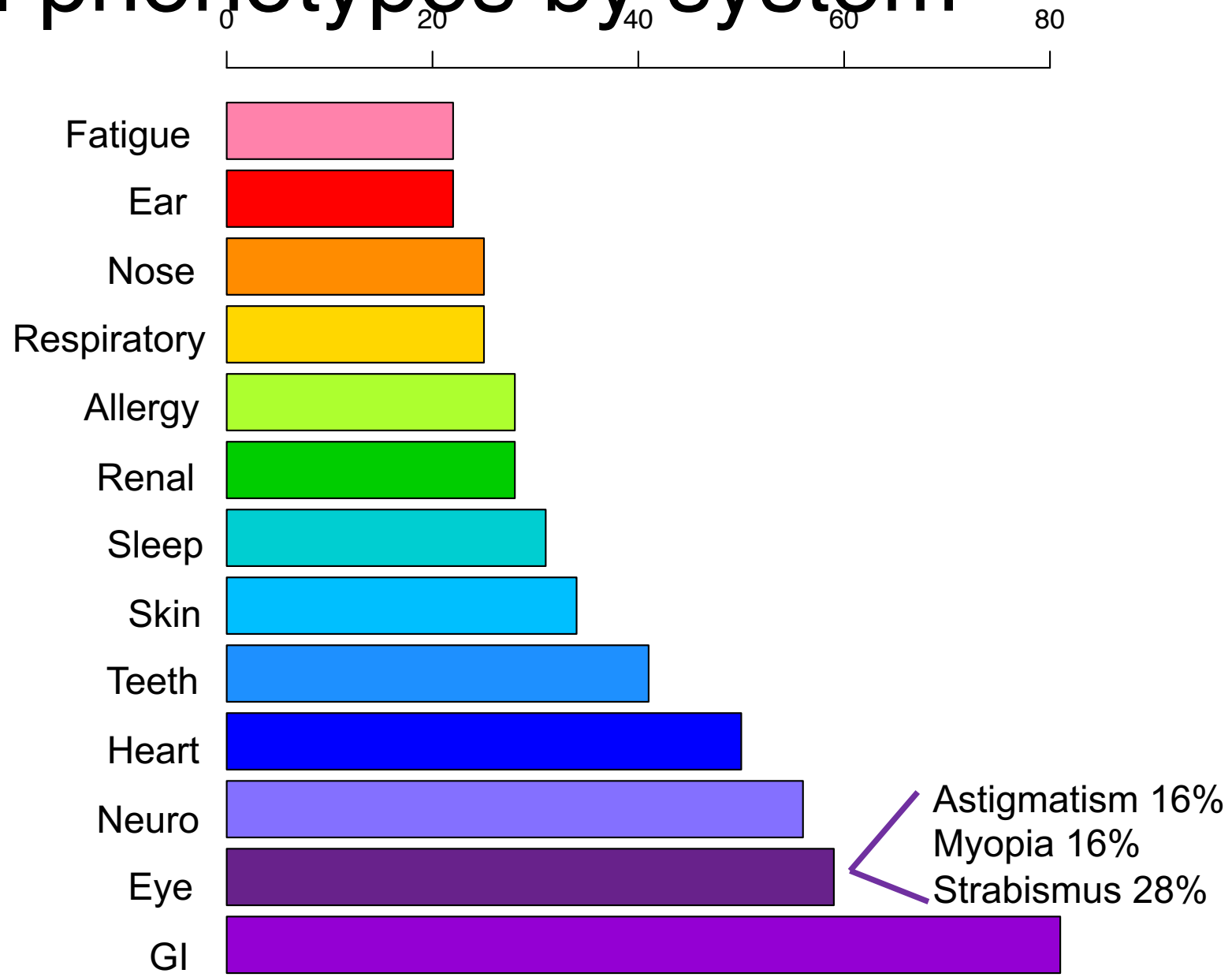
Medical phenotypes by system



Feeding problems in infancy 59%
Reflux 50%
Feeding problems beyond infancy 16%
Failure to thrive beyond infancy 41%
Constipation 41%

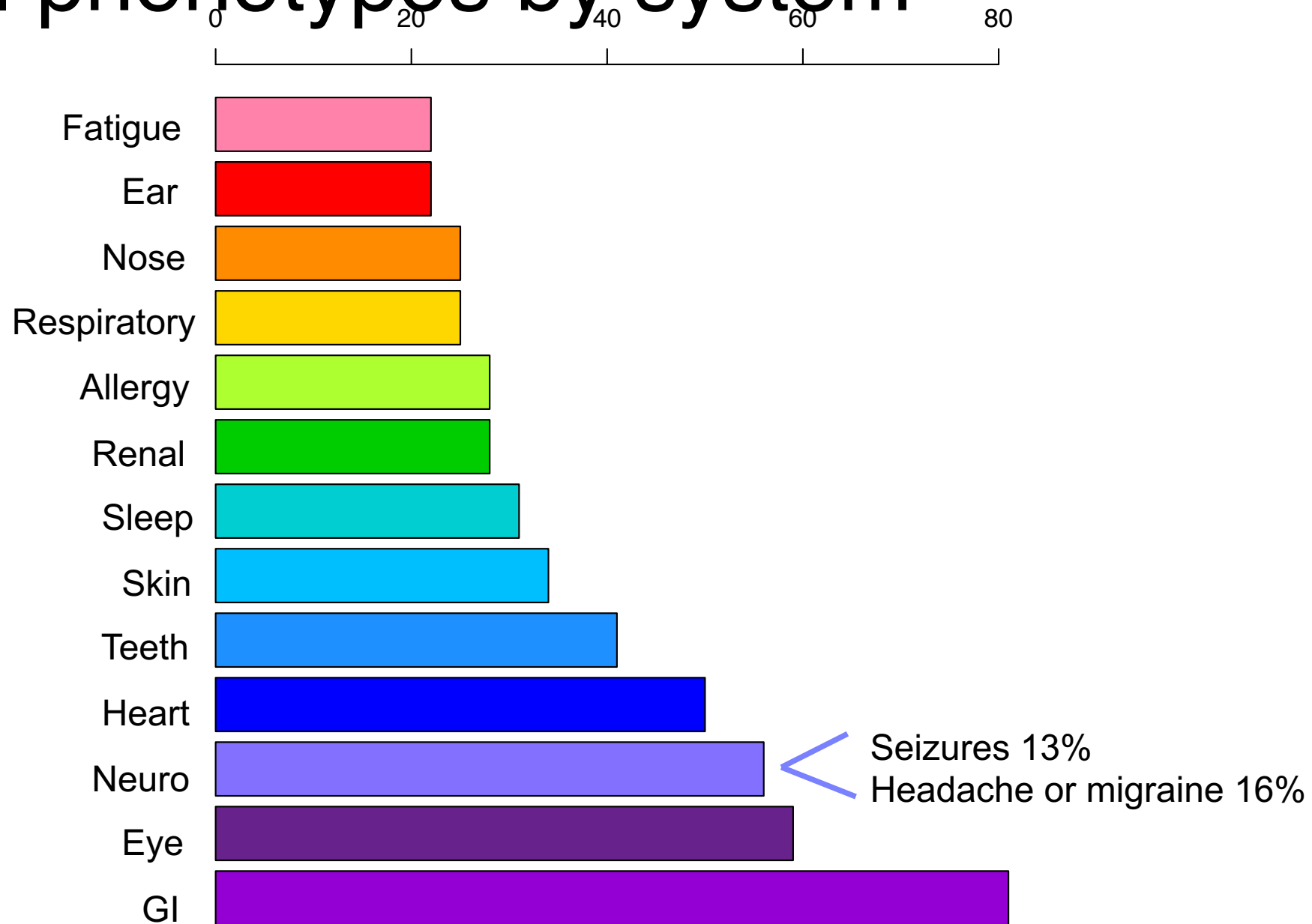
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Medical phenotypes by system



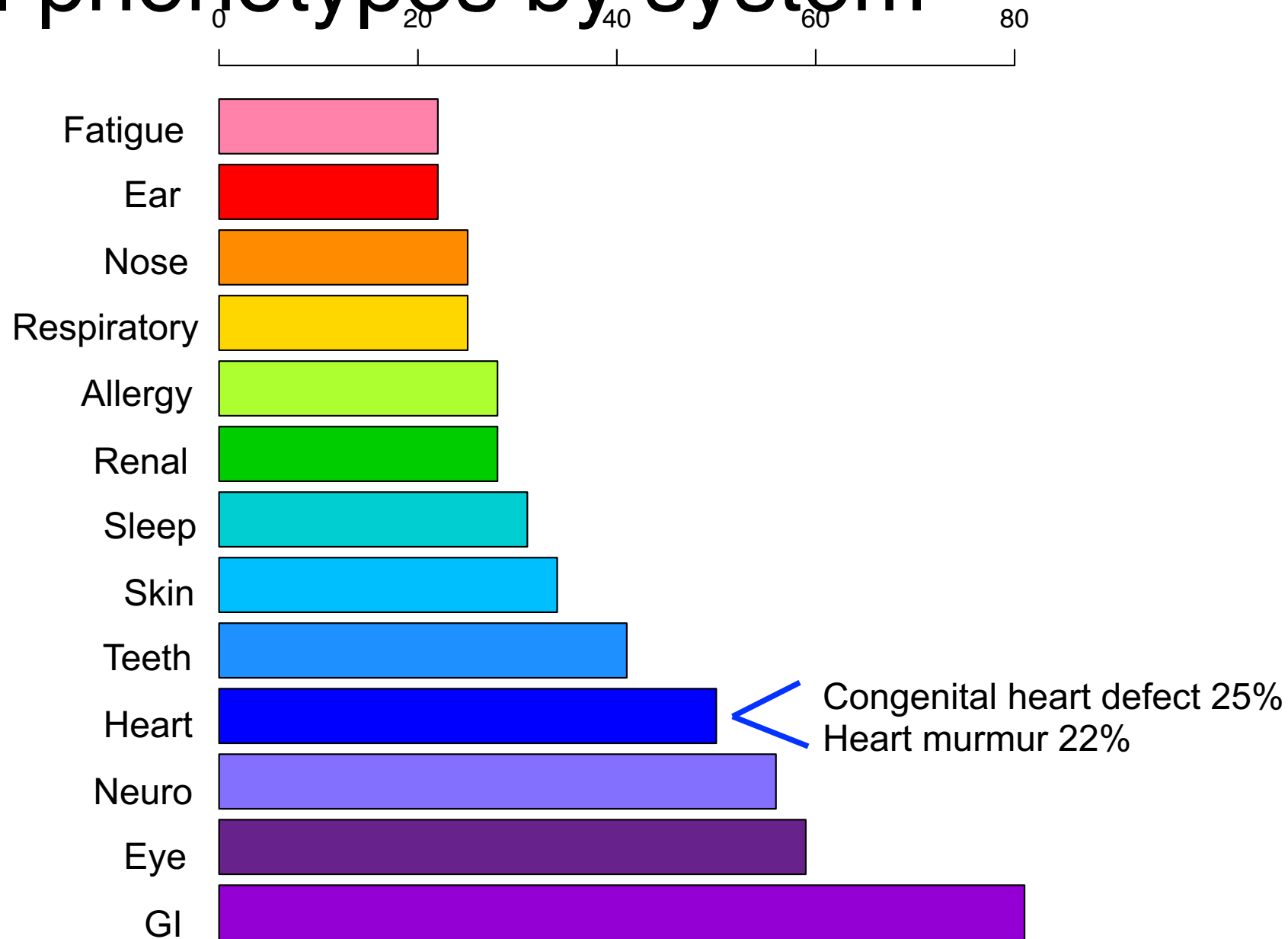
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Medical phenotypes by system



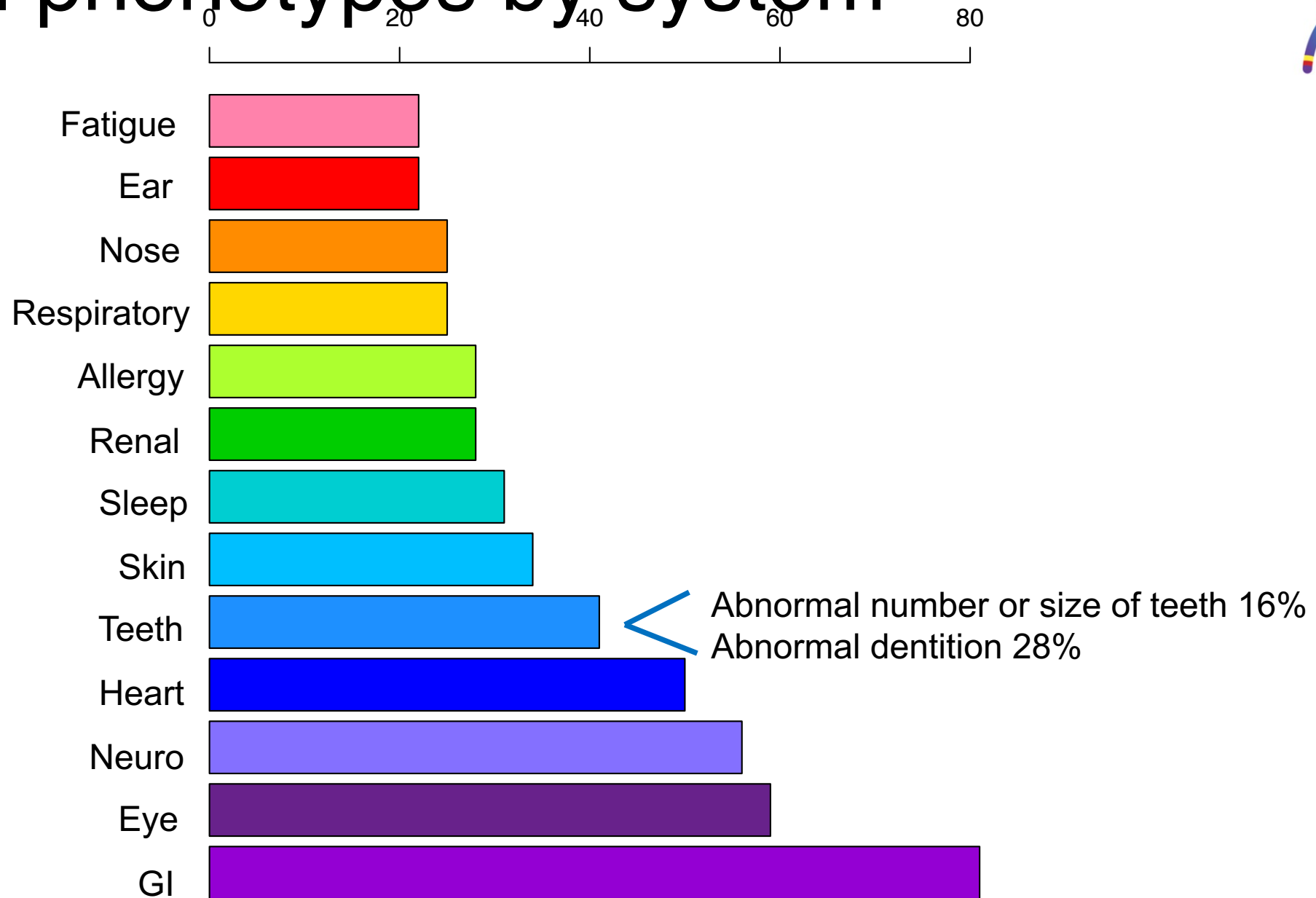
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Medical phenotypes by system



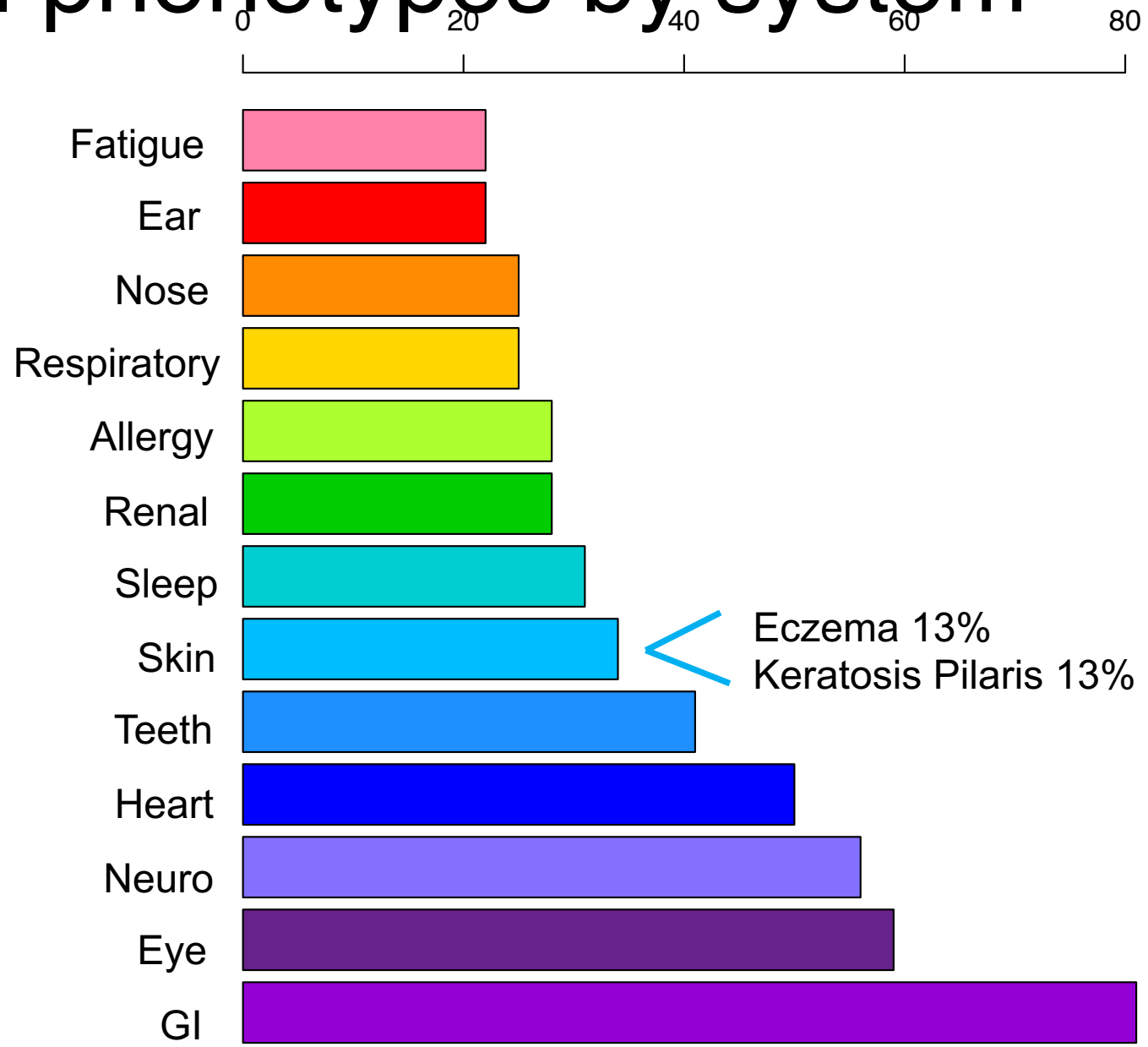
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Medical phenotypes by system



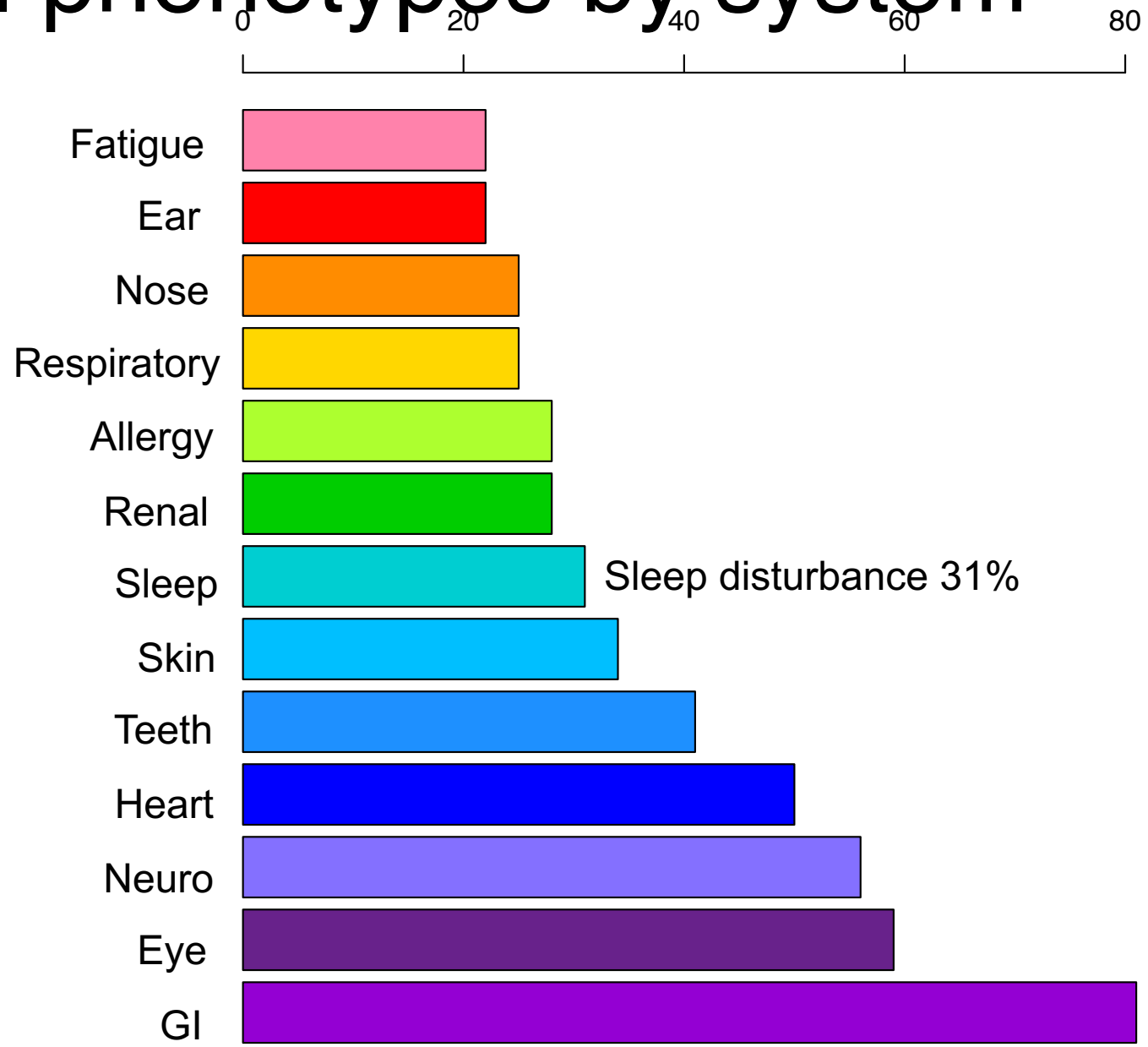
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Medical phenotypes by system



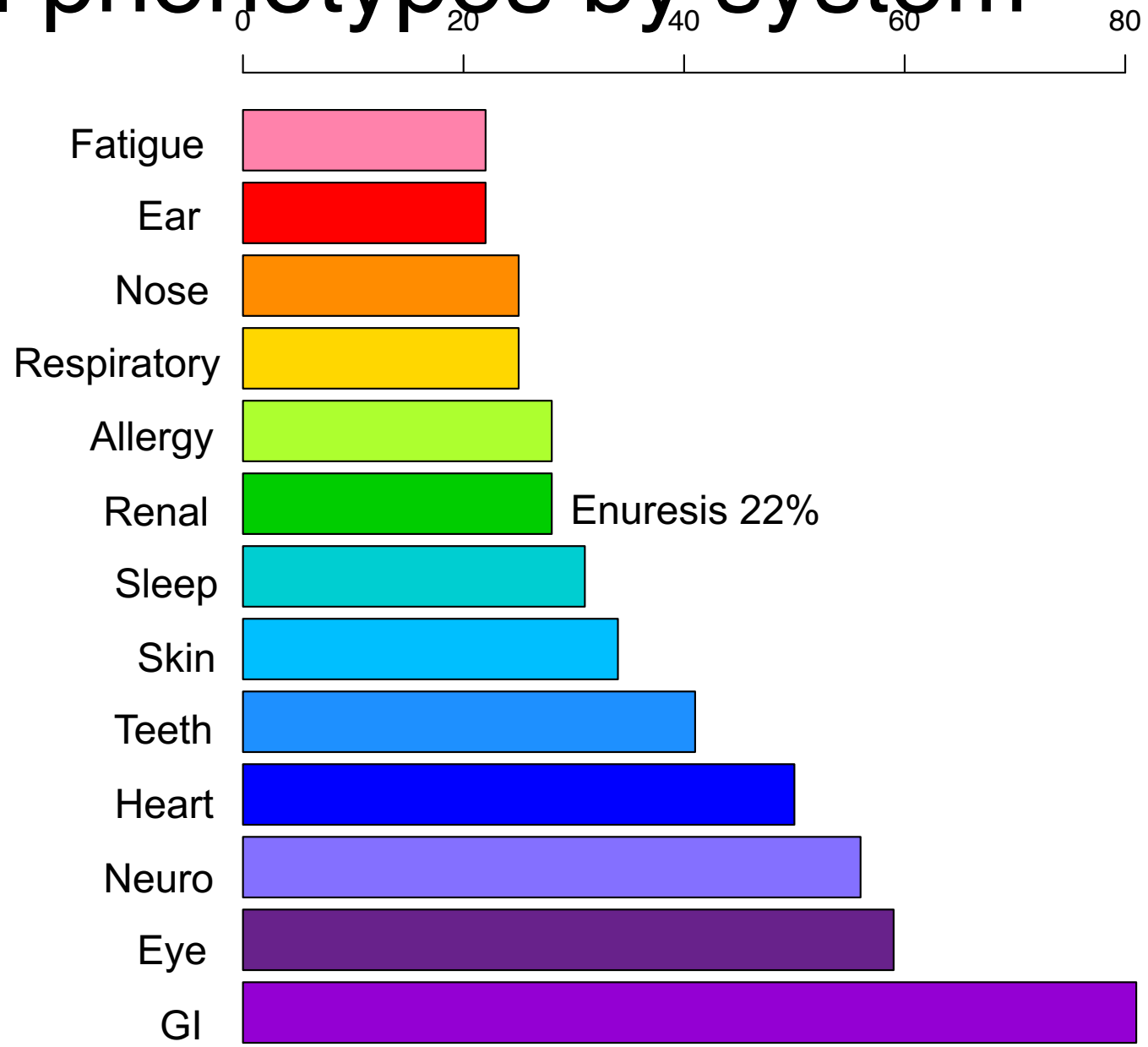
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Medical phenotypes by system



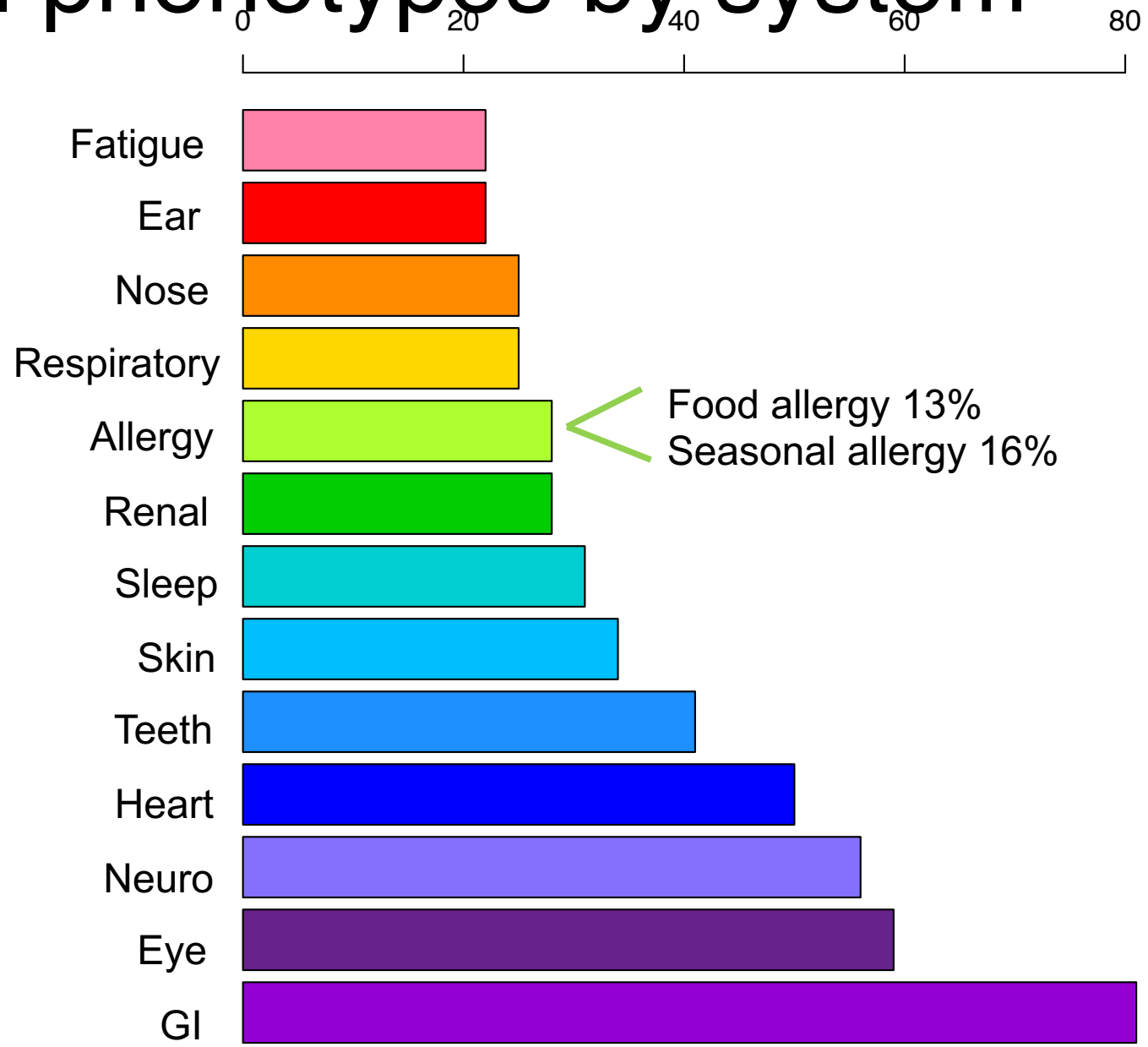
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Medical phenotypes by system



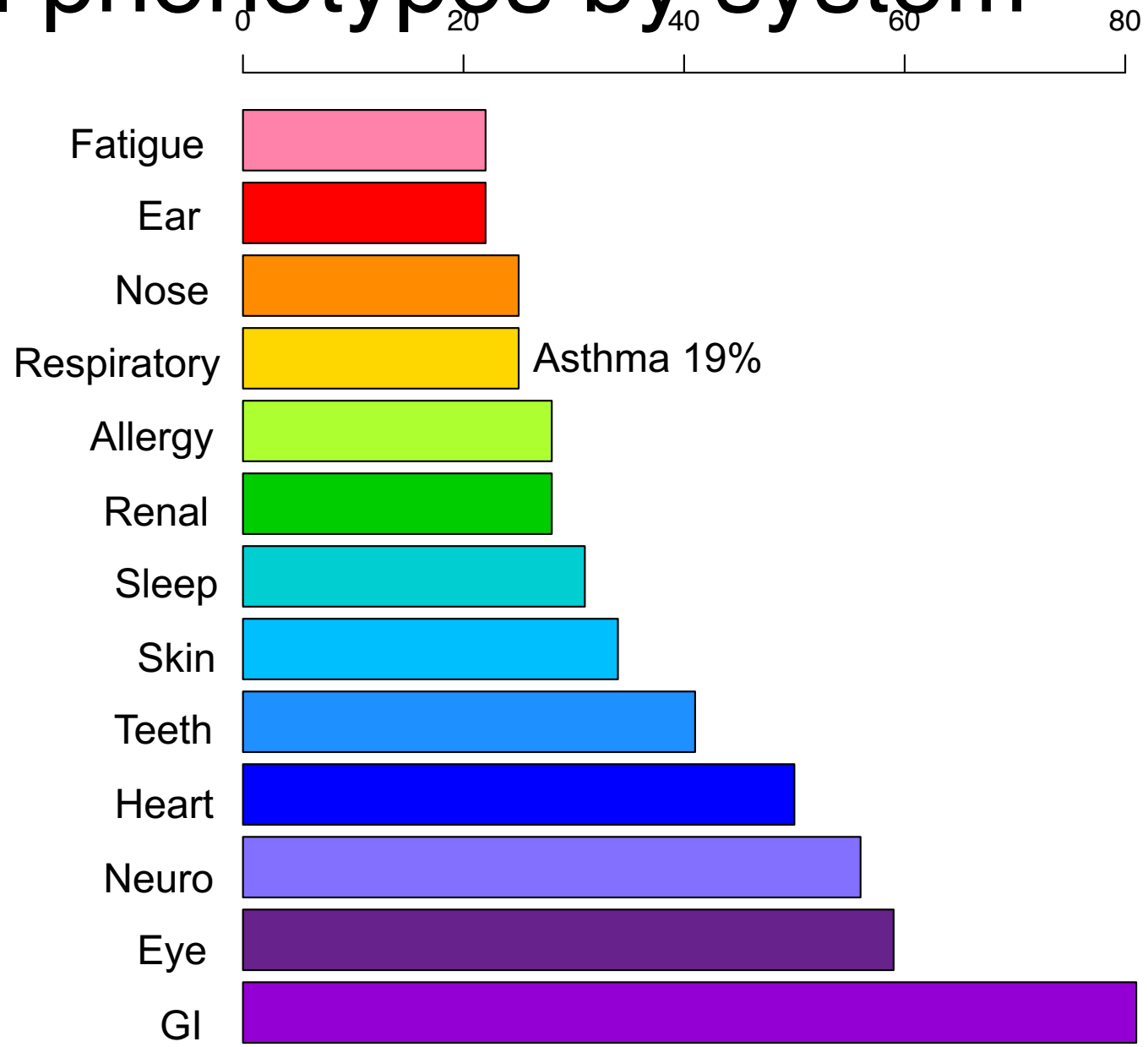
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Medical phenotypes by system



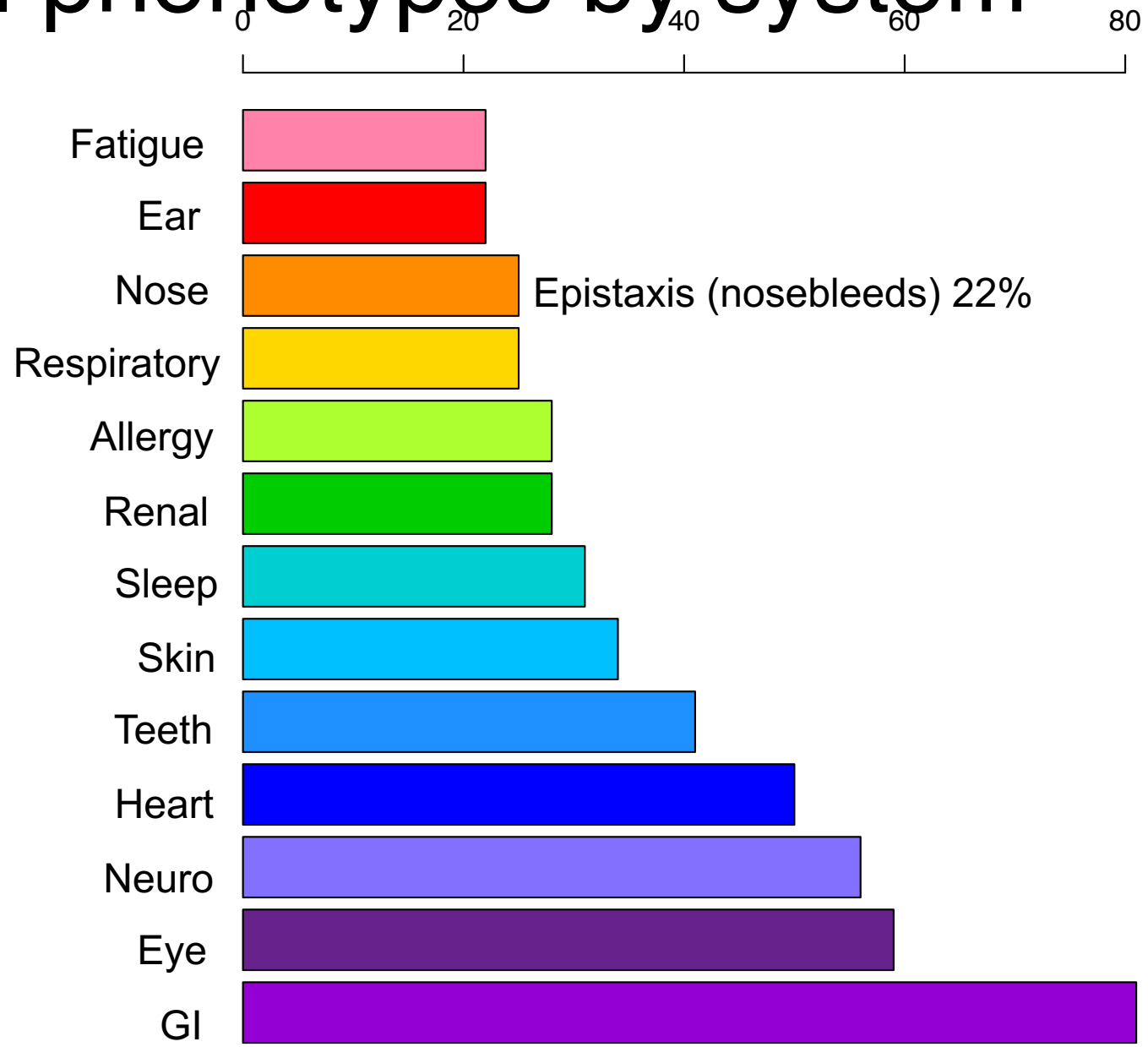
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Medical phenotypes by system



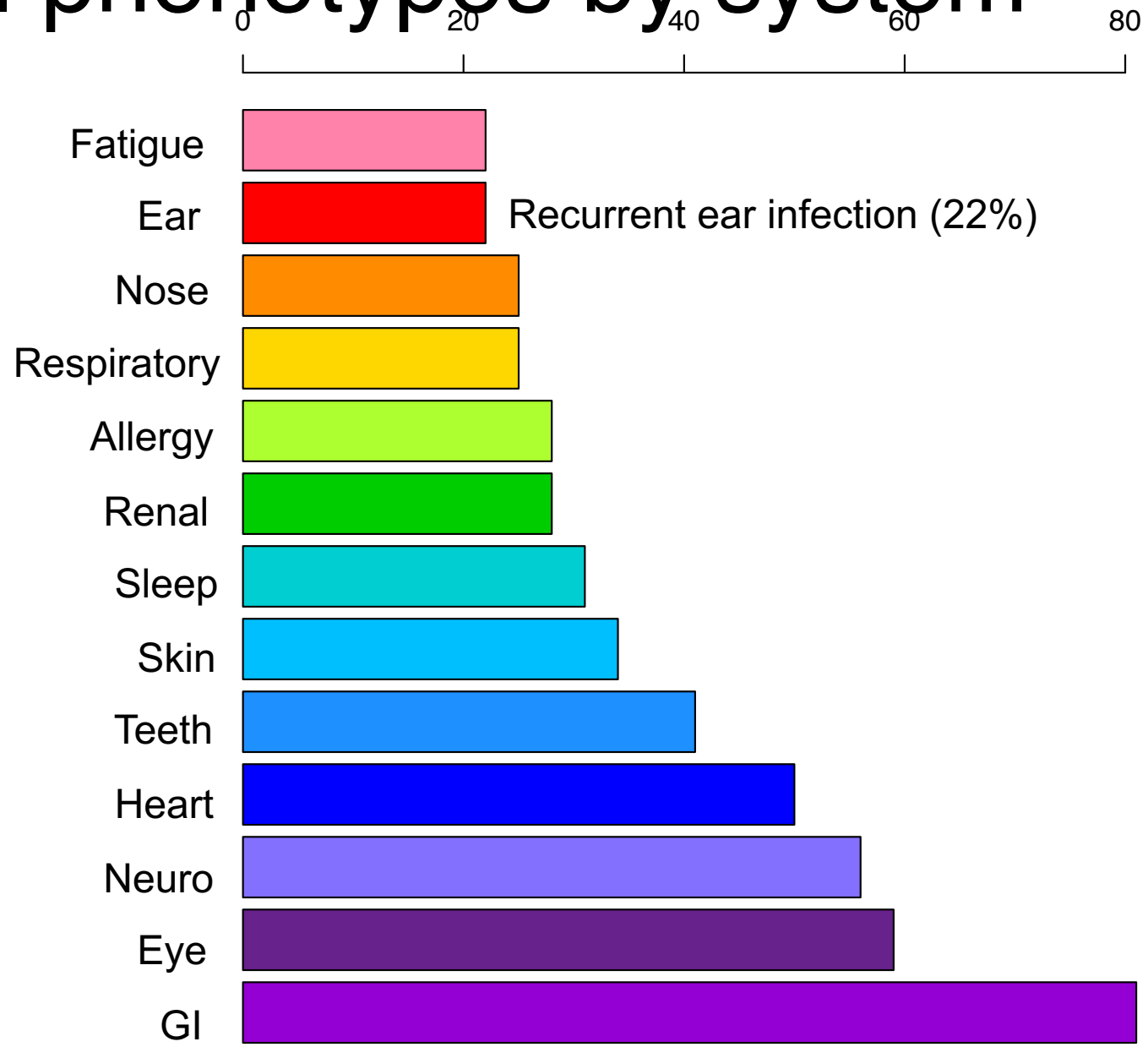
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Medical phenotypes by system



00:25:54

Medical phenotypes by system



00:26:50

Question and Answer Session