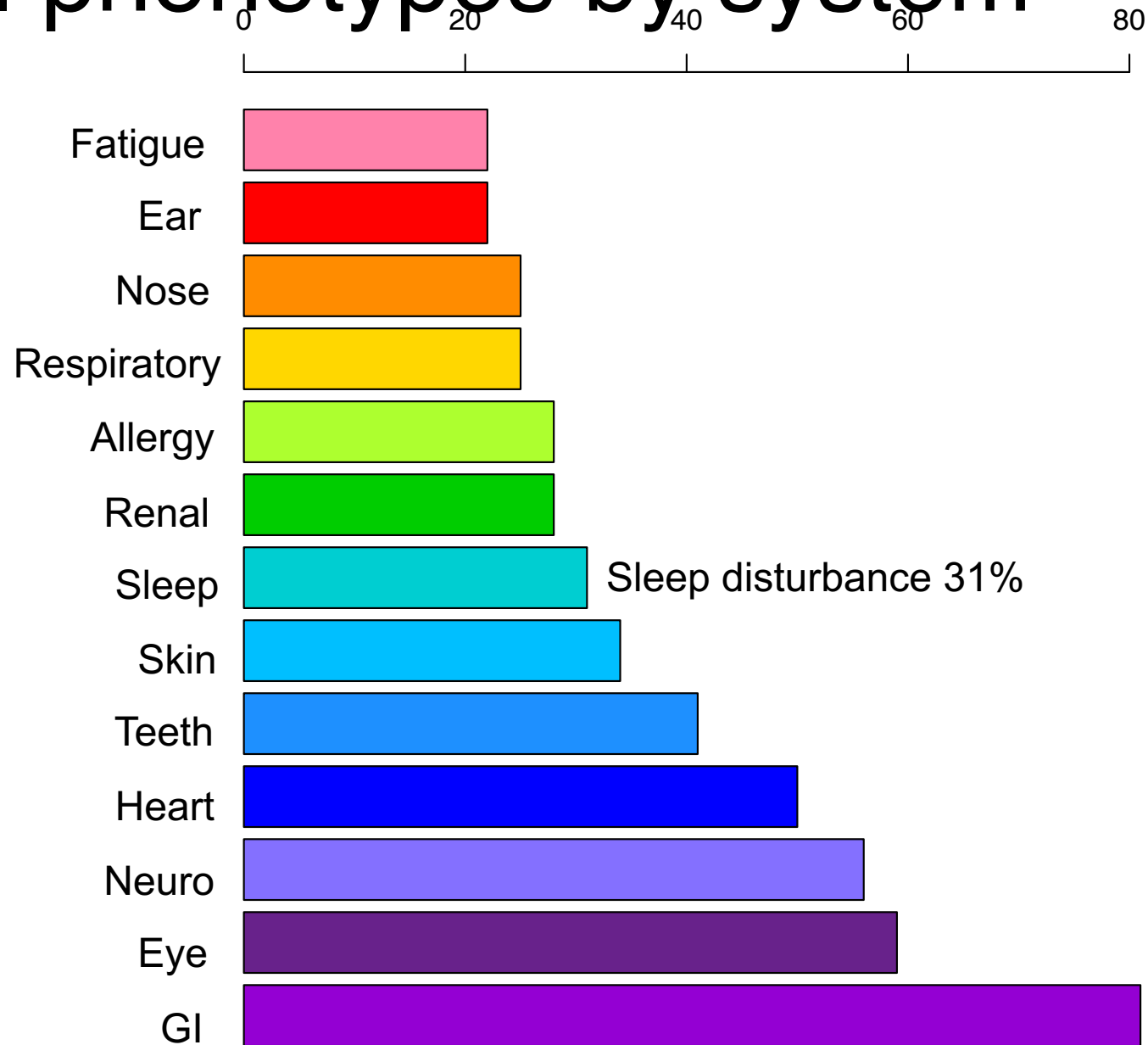
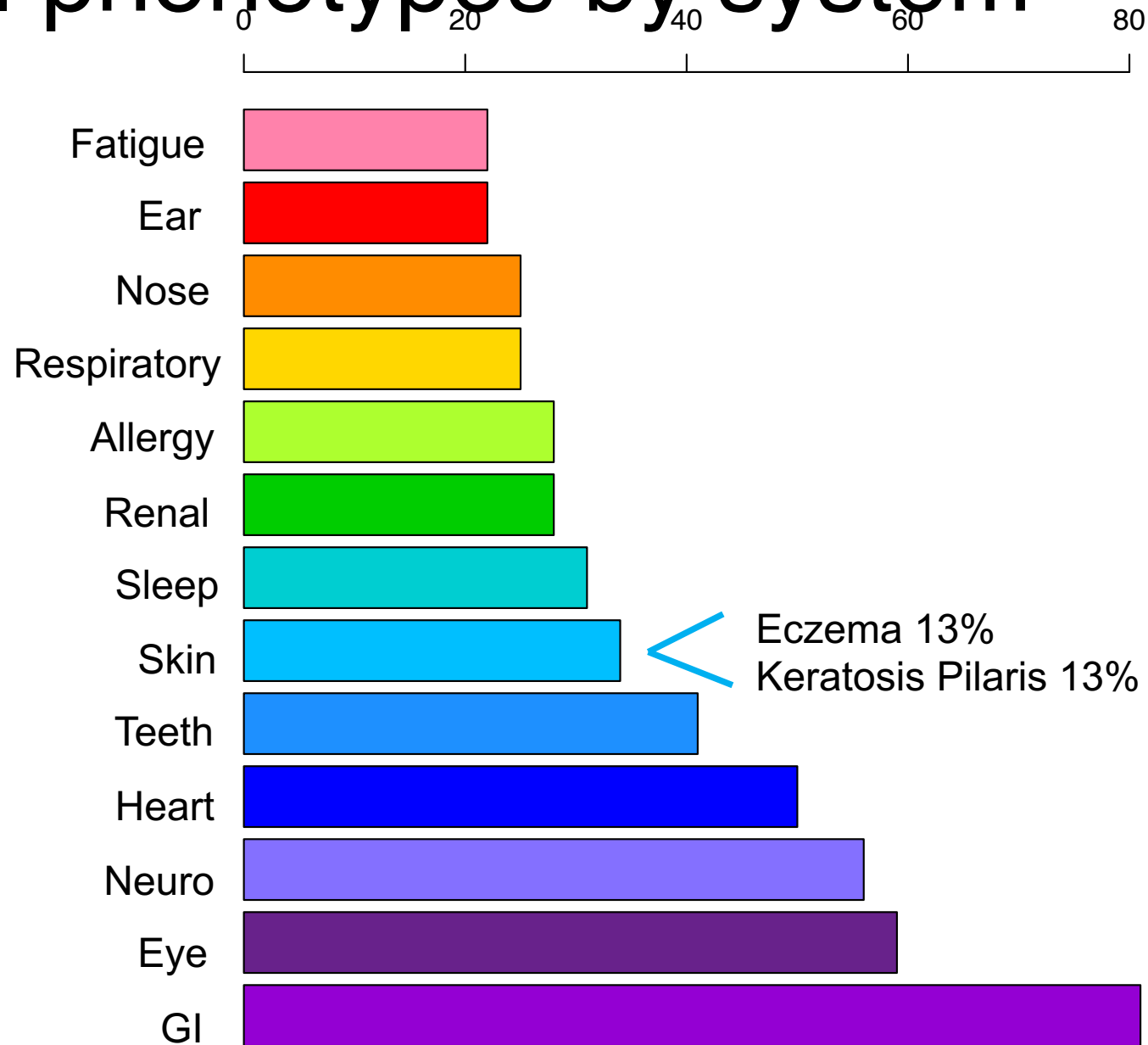


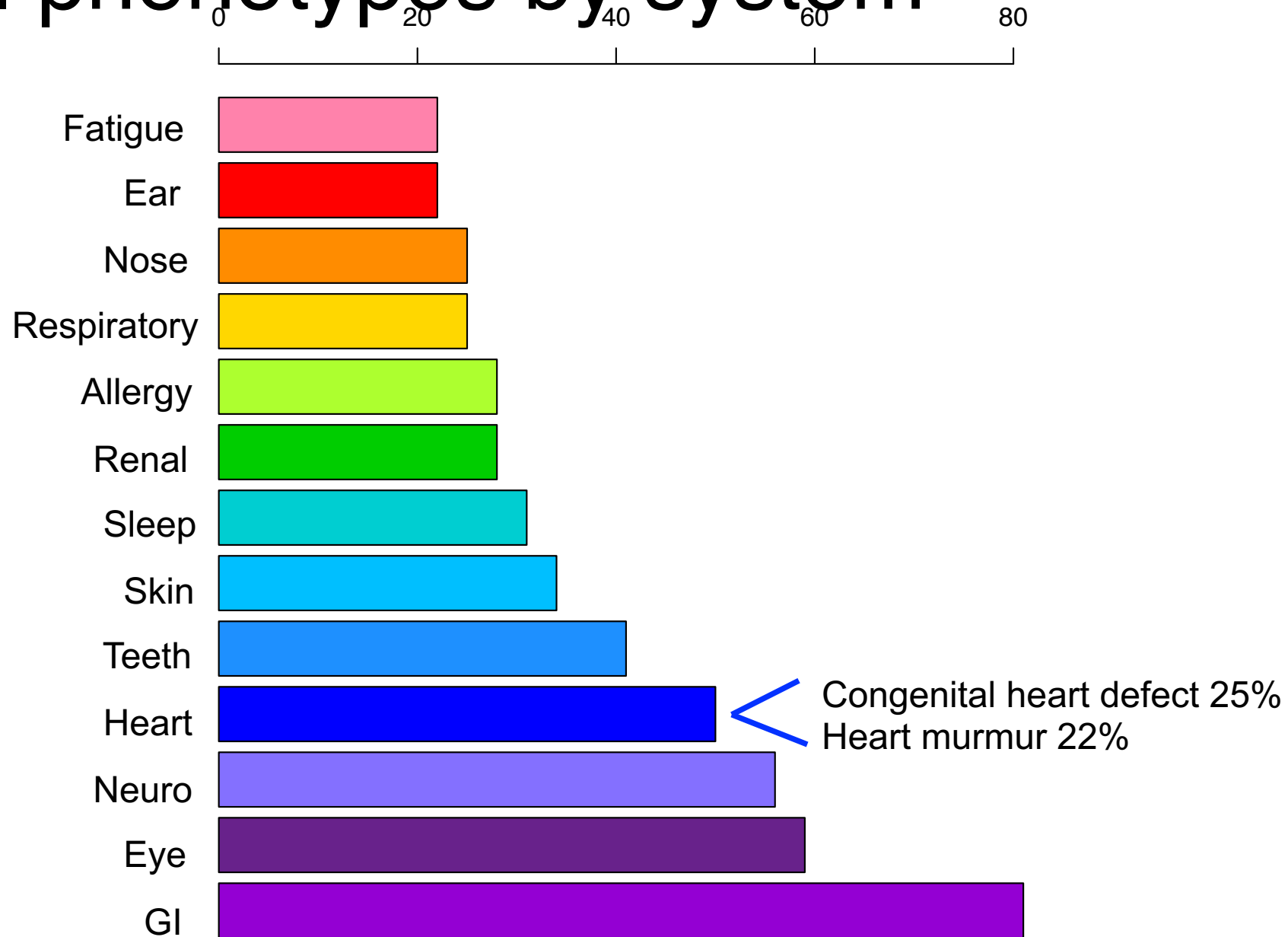
# Medical phenotypes by system



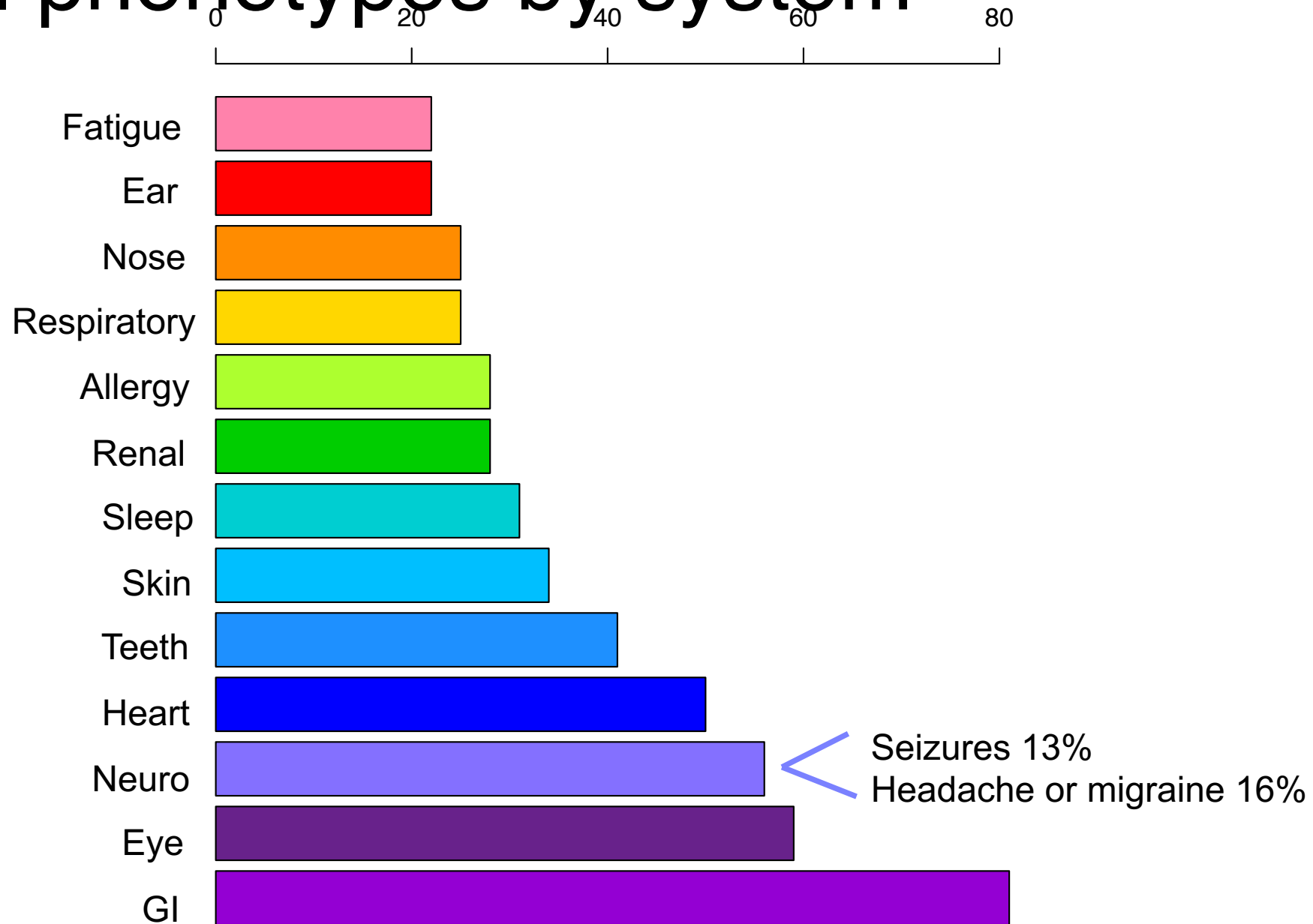
# Medical phenotypes by system



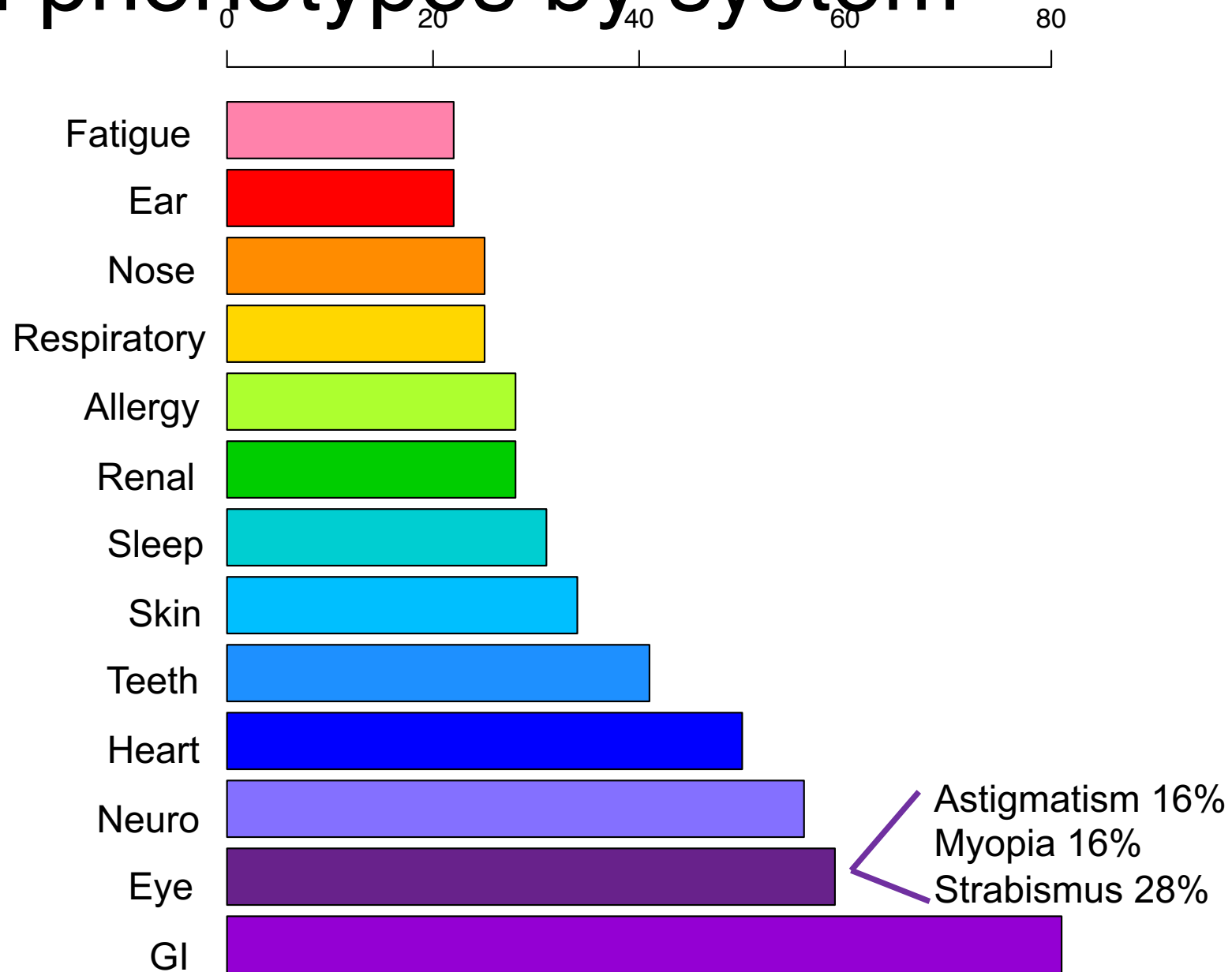
# Medical phenotypes by system



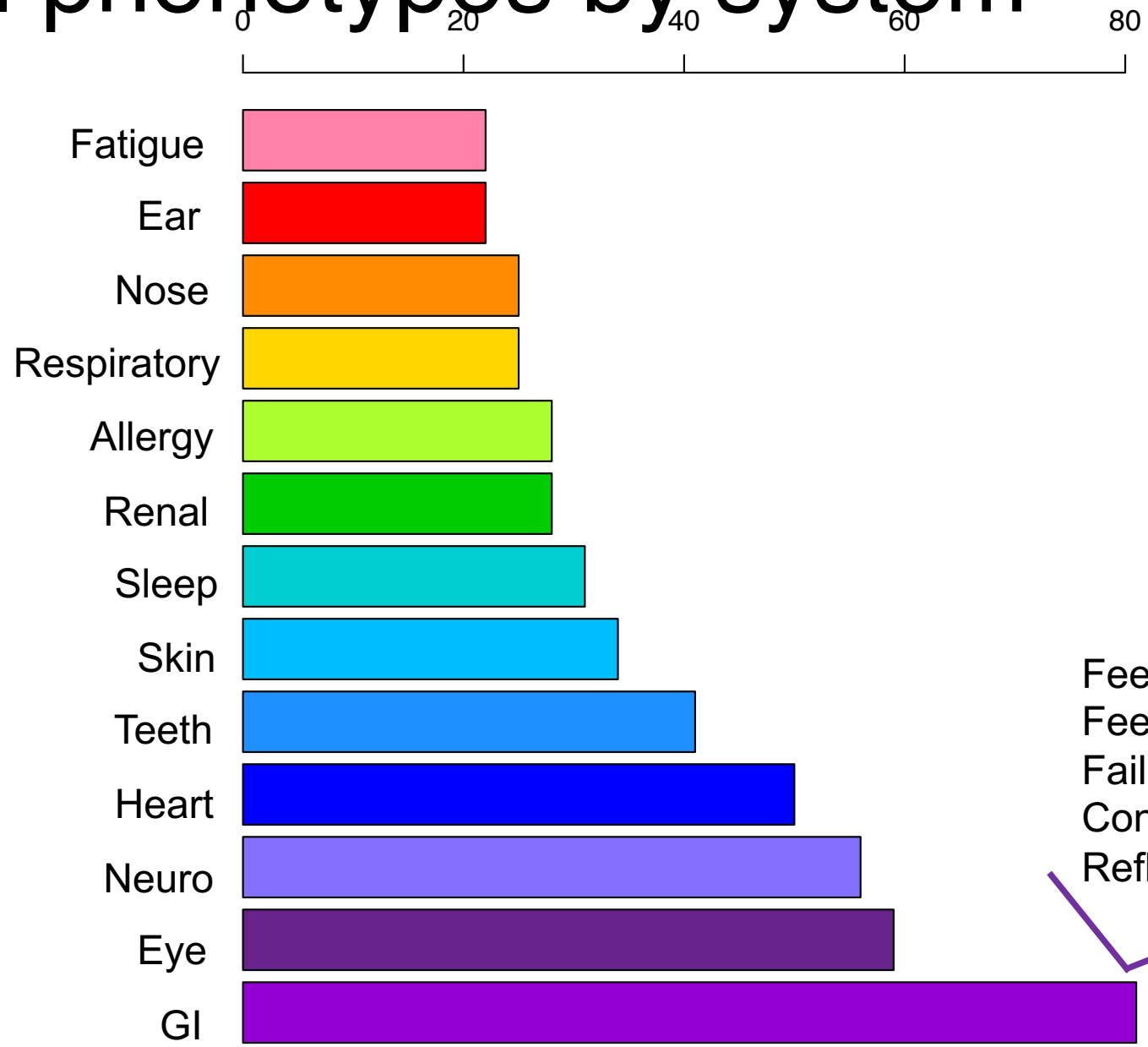
# Medical phenotypes by system



# Medical phenotypes by system

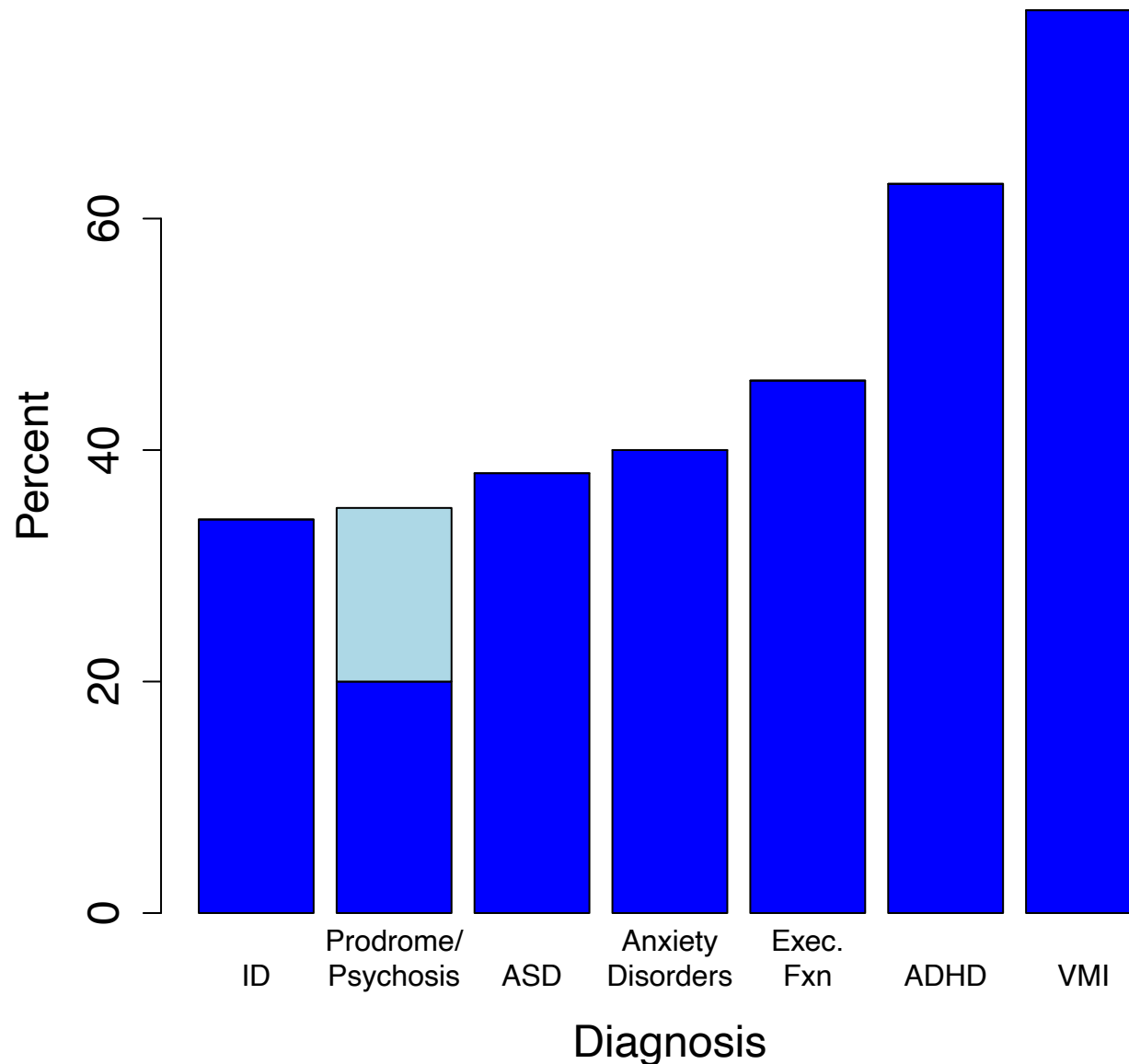


# Medical phenotypes by system



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

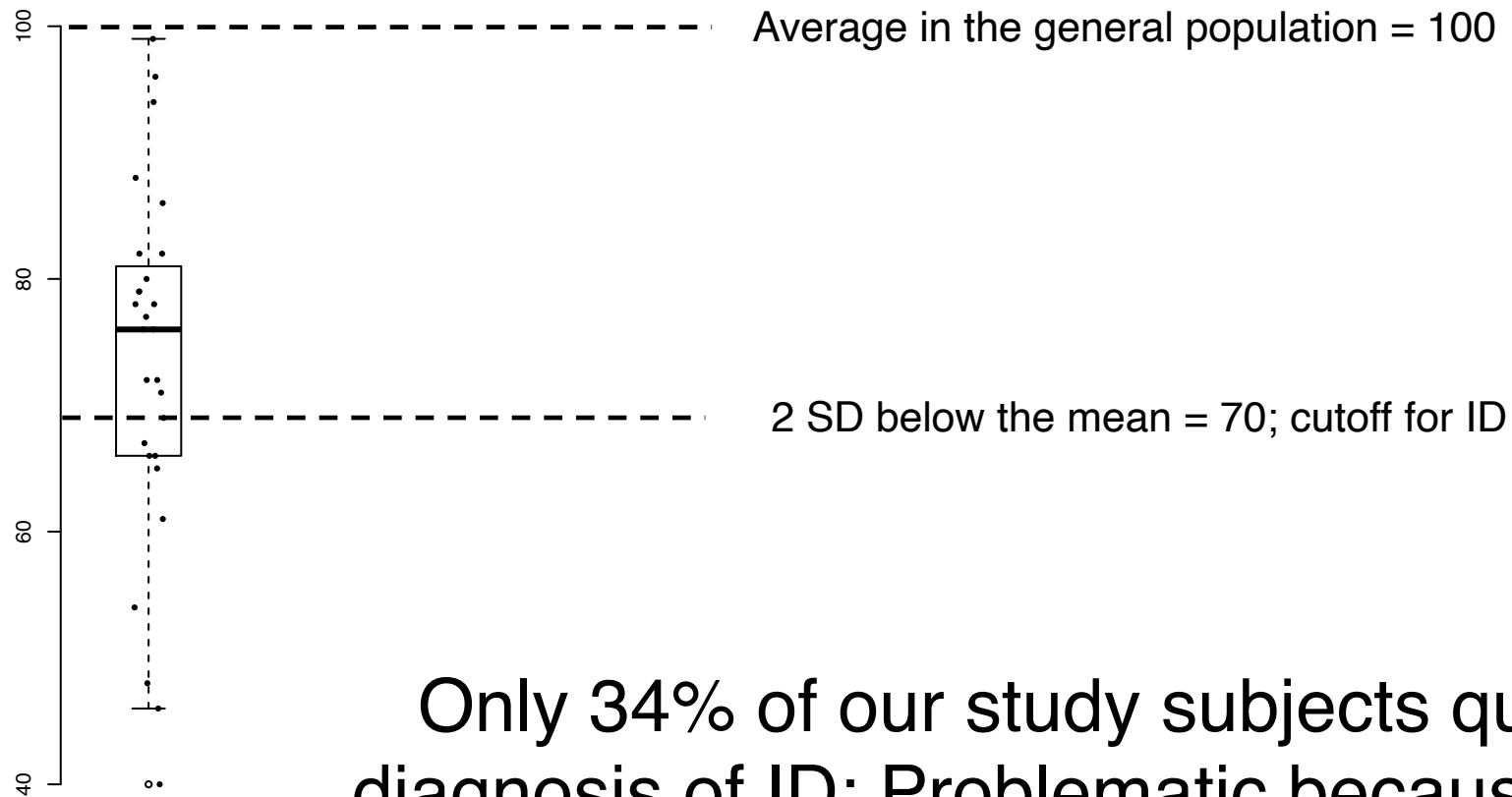
# Neurodevelopmental and Neuropsychiatric Diagnoses



Anxiety includes Generalized Anxiety Disorder, Specific Phobia, Separation Anxiety, Social Anxiety

# Cognitive Disability

Full-Scale IQ:



Avg IQ = 73  
Range 40-99

Only 34% of our study subjects qualify for a diagnosis of ID: Problematic because access to many services is tied to diagnosis of ID



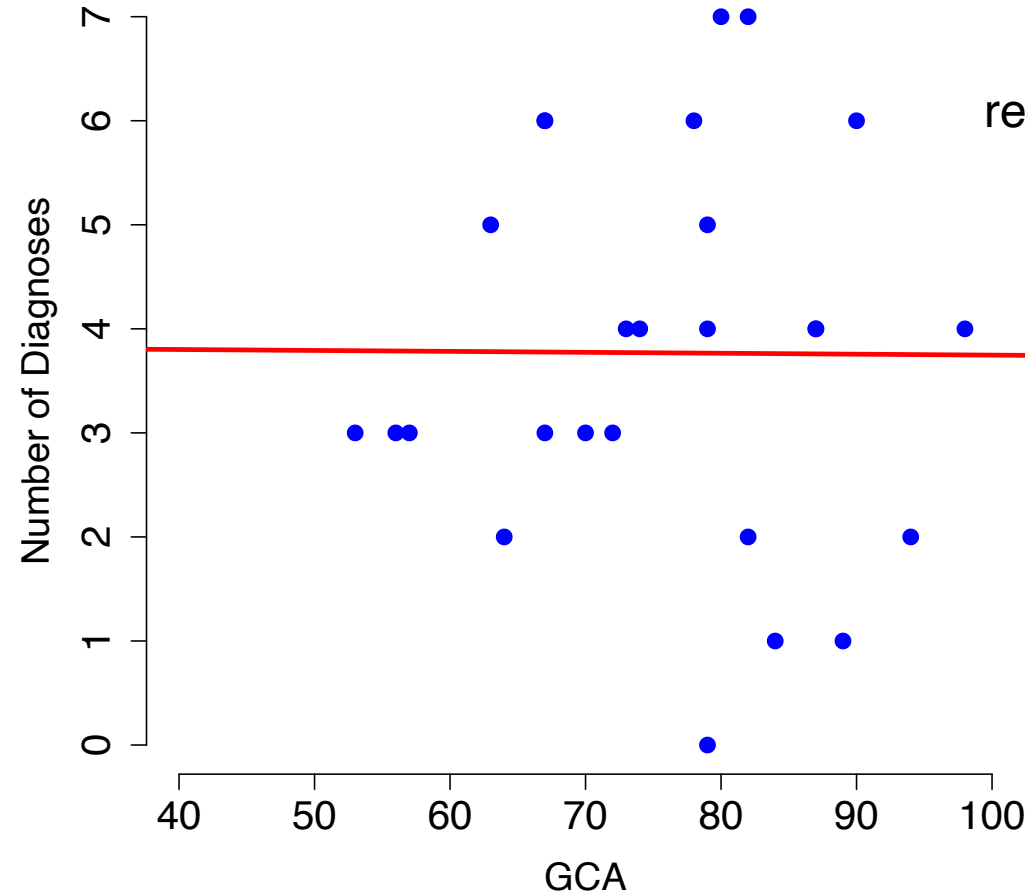
# Cognitive Disability vs Overall Disability



Possible diagnoses:

- GAD
- Separation anxiety
- Social anxiety
- Specific phobia
- ASD
- ADHD
- Adaptive behavior
- Conduct disorder
- Enuresis
- Executive function
- Graphomotor weakness
- ODD
- Psychosis

Possible range 0 – 13  
Actual Range 0-7



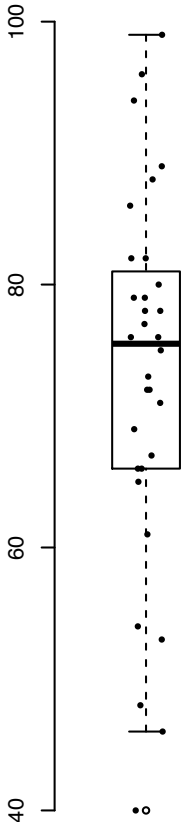
Slope of regression line = -0.001  
p value = 0.97

**Measures of cognitive ability, while important in their own right, are not a useful proxy for overall disability in 3q29 deletion individuals.**

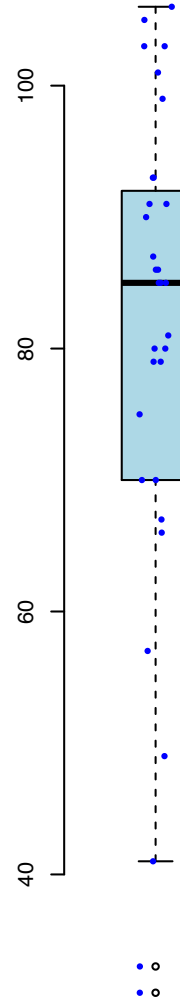
# Components of Intellectual Function



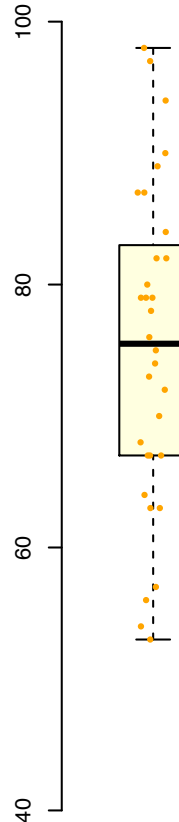
FSIQ  
Avg = 73  
Range 40-99



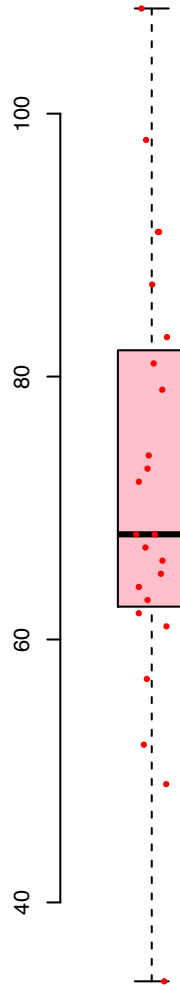
Verbal  
Avg = 80  
Range 31-106



Nonverbal Reasoning  
Avg = 75  
Range 53-98

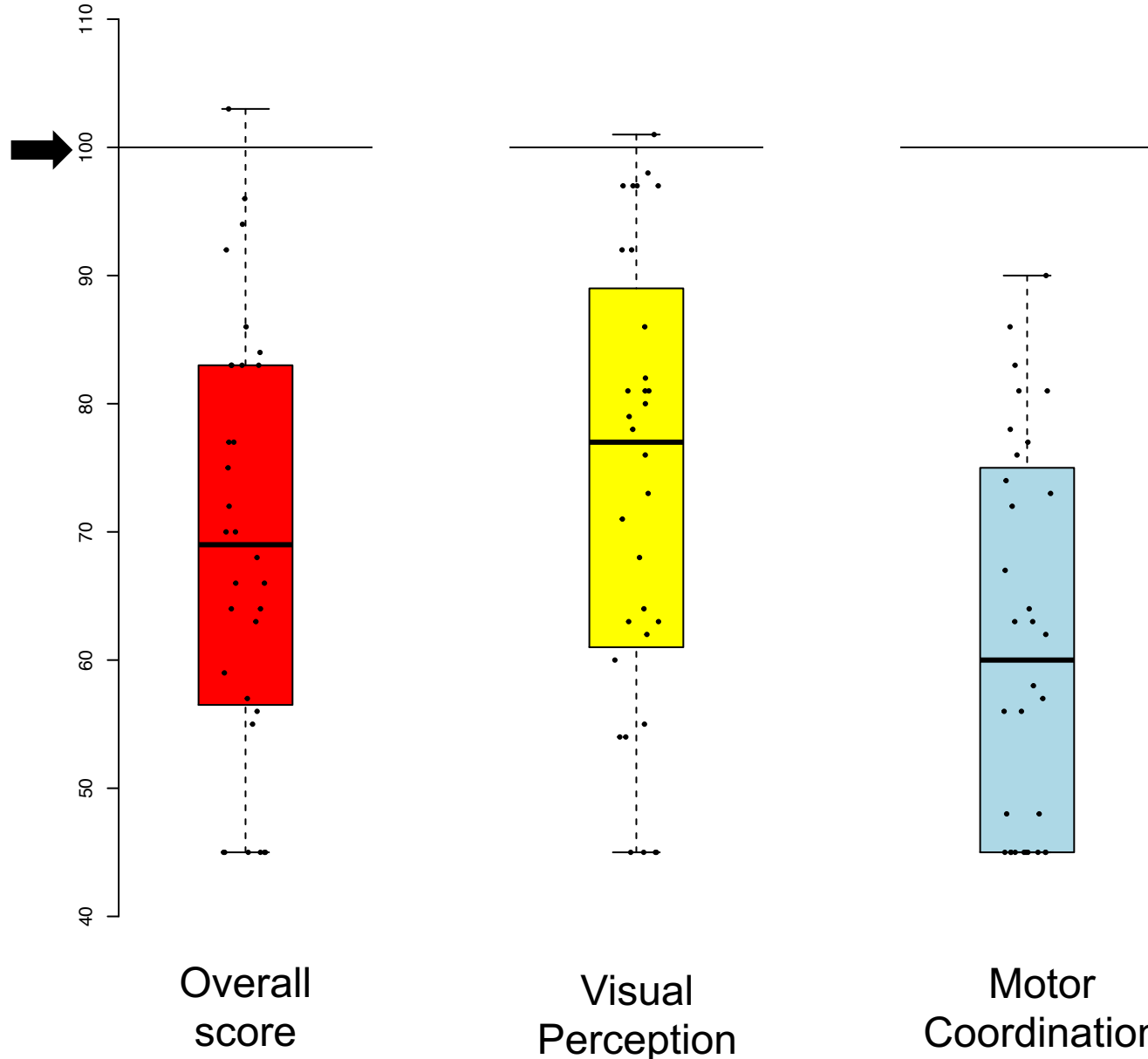


Spatial Reasoning  
Avg = 71  
Range 34-108



# Graphomotor weakness

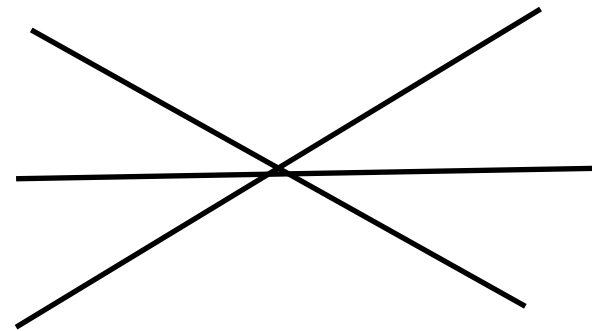
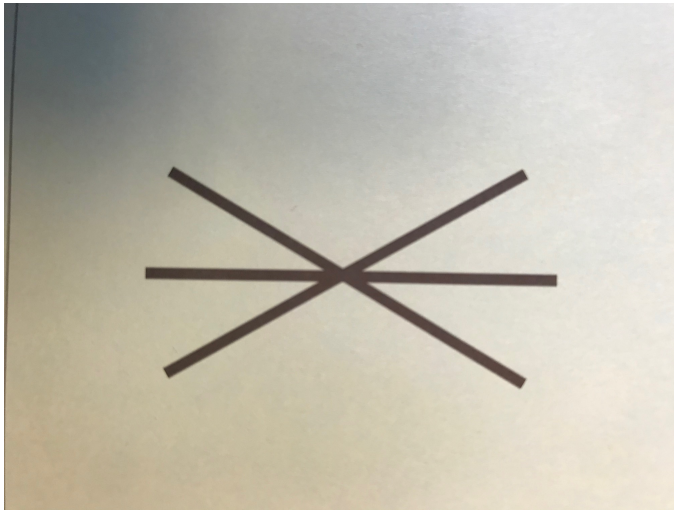
Expected  
score in  
the  
general  
population



These data indicate that there is pronounced graphomotor weakness in individuals with the 3q29 deletion, with a substantial deficit in motor coordination

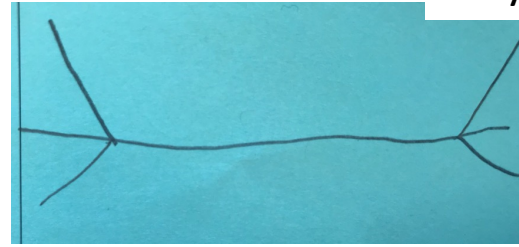
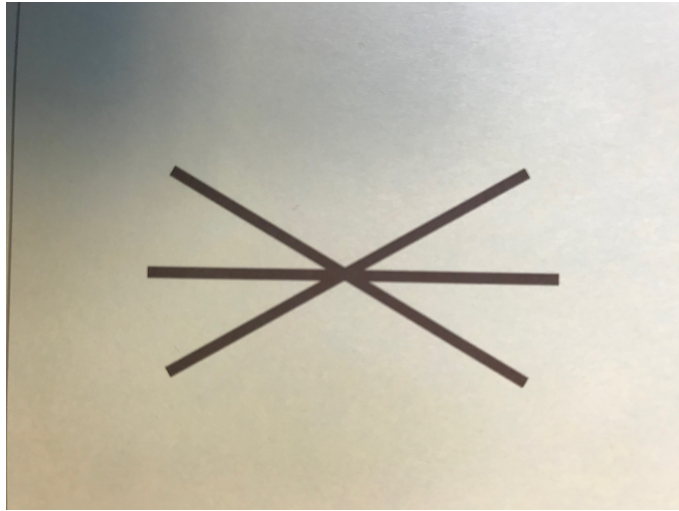
# Visual-motor integration

Draw this:

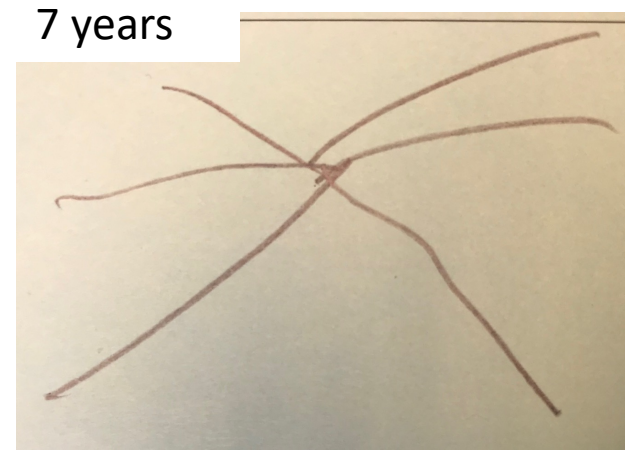


# Visual-motor integration

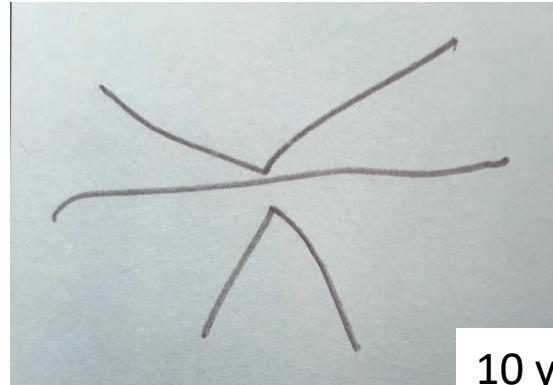
Draw this:



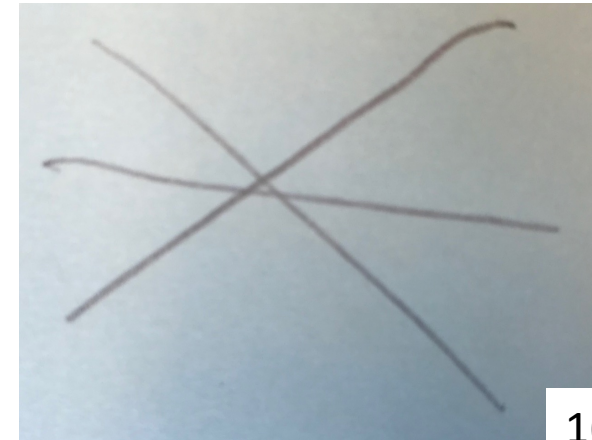
15 years



7 years

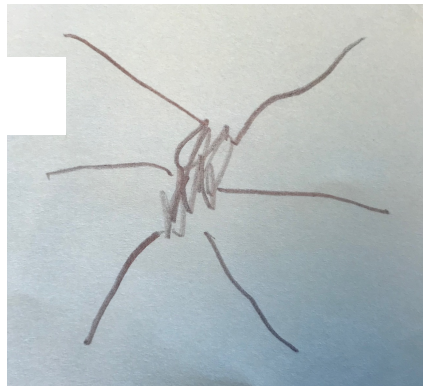


10 years

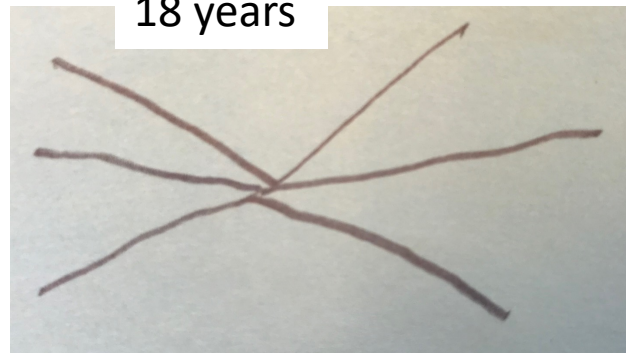


16 years

6 years



18 years



18 years