- GROUPS….CONFRIM WEBSITE

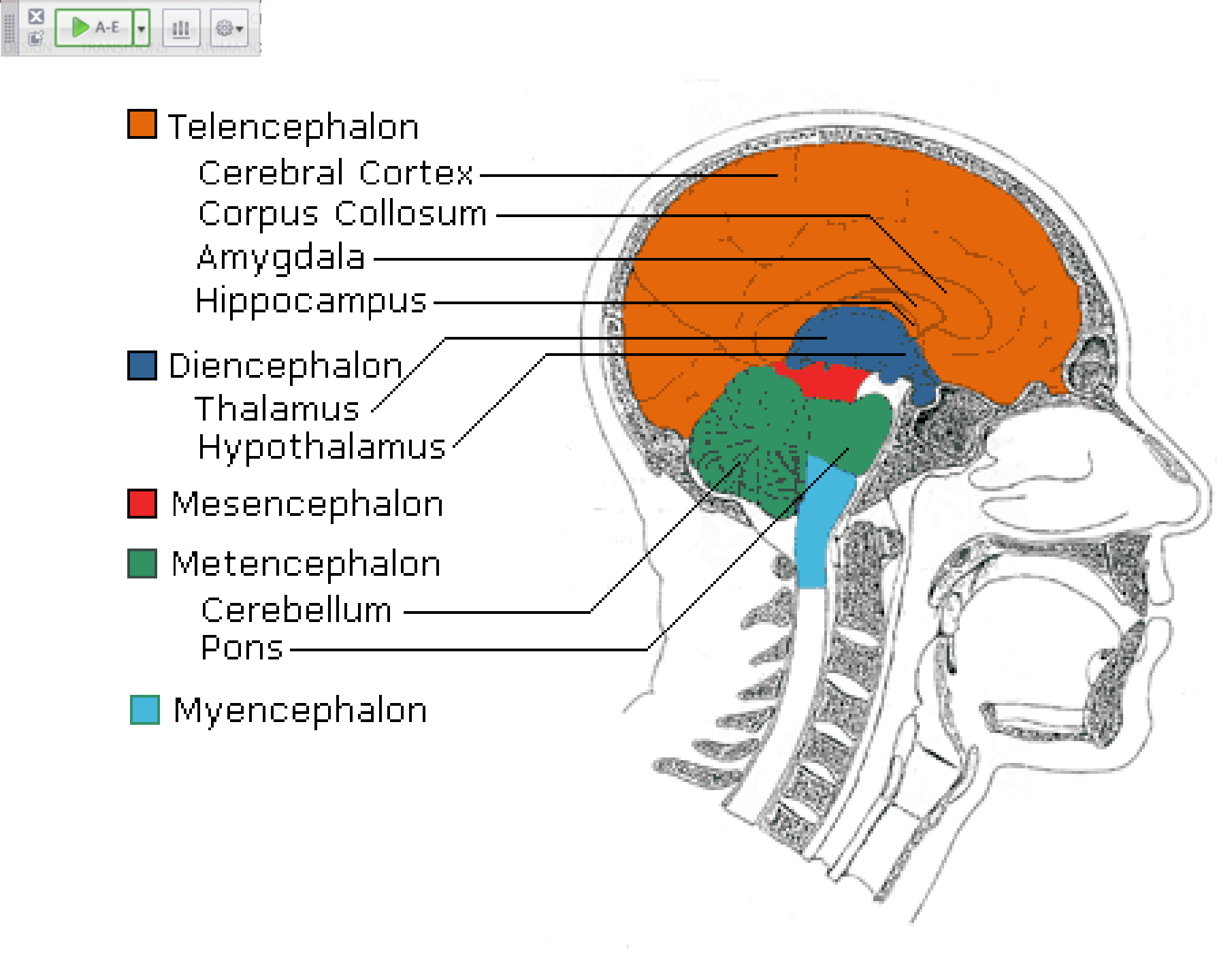
Autism Spectrum Disorder (ASD)

- ~1% American adults
- Higher recent estimates (1 in 50 births)
- Males 4x more likely
- Familial association
- Possible associated with older parents
- Savant phenotypes ~10%

ASD Drug Treatments

NO drugs for specifically treating ASD

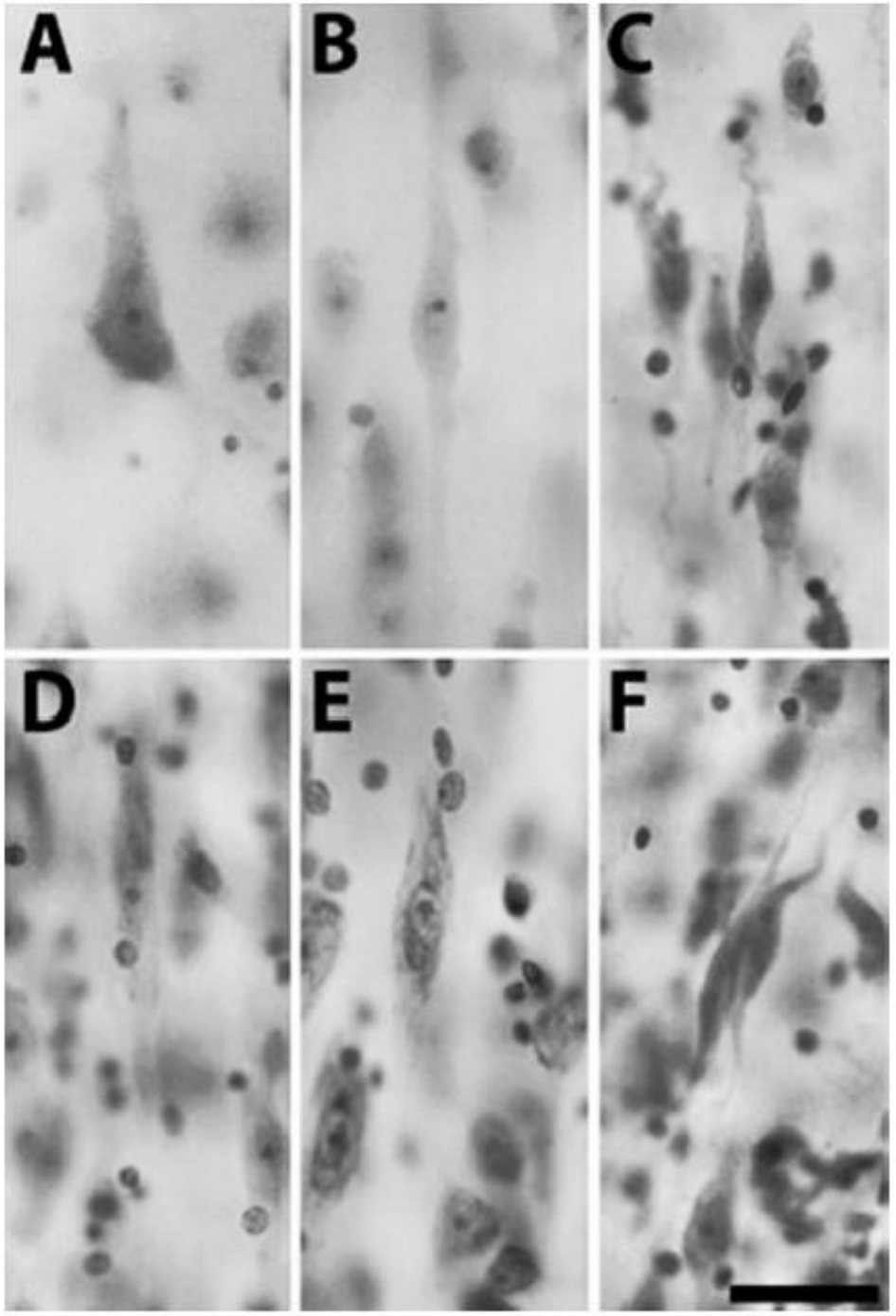
Antipsychotics and Antidepressants sometime prescribed for symptoms

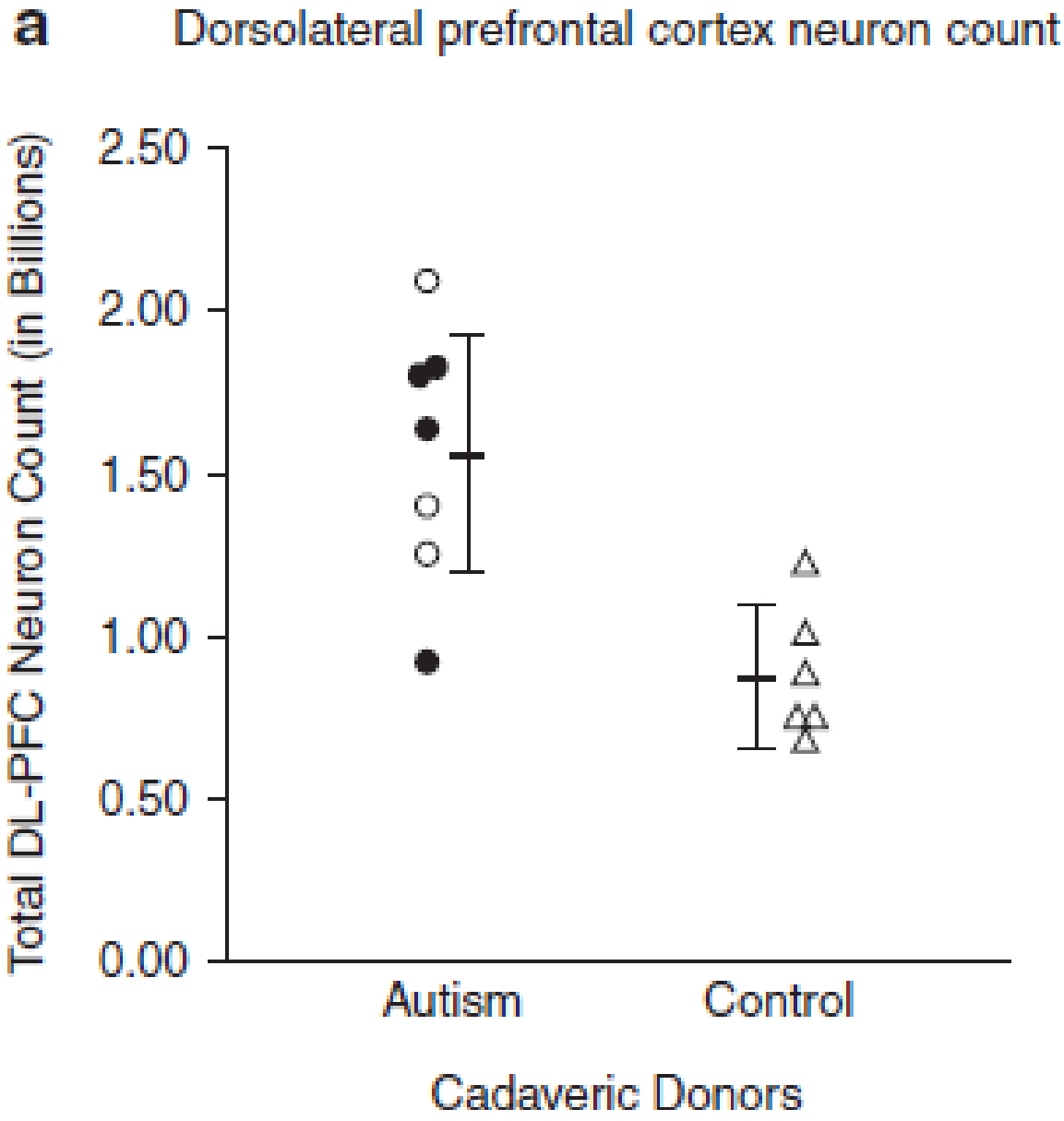

Telencephalon
Cerebral Cortex
Corpus Collosum
Amygdala
Hippocampus
Diencephalon
Thalamus
Hypothalamus
Mesencephalon
Metencephalon
Cerebellum
Pons
Myencephalon

Cell counting using these images is

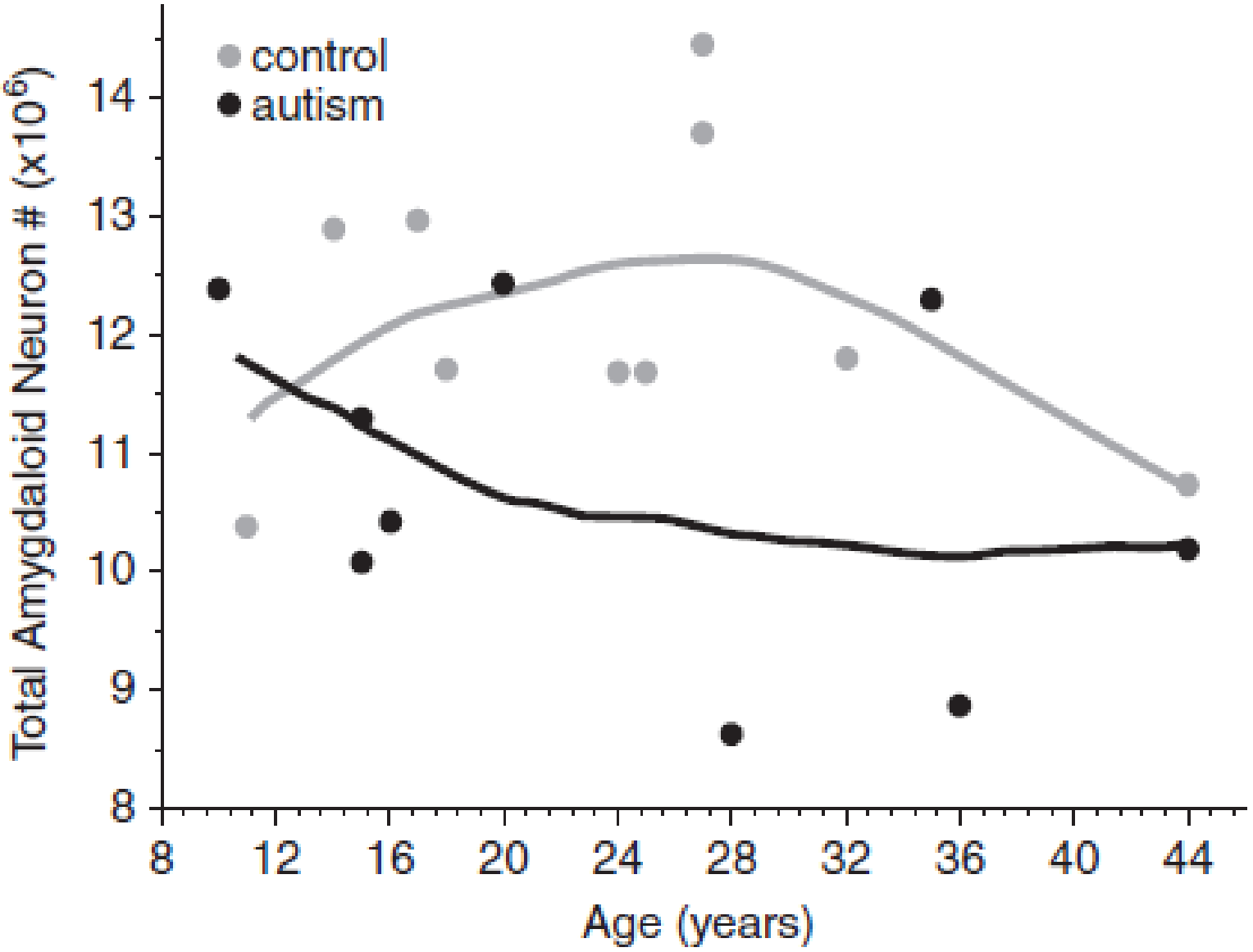
A. Objective
B. Somewhat subjective
C. Error free
D. A and C

WHY?




a
Dorsolateral prefrontal cortex neuron count
Total DL-PFC Neuron Count (in Billions)
2.50
2.00
1.50
1.00
0.50
0.00
Autism
Control
Cadaveric Donors

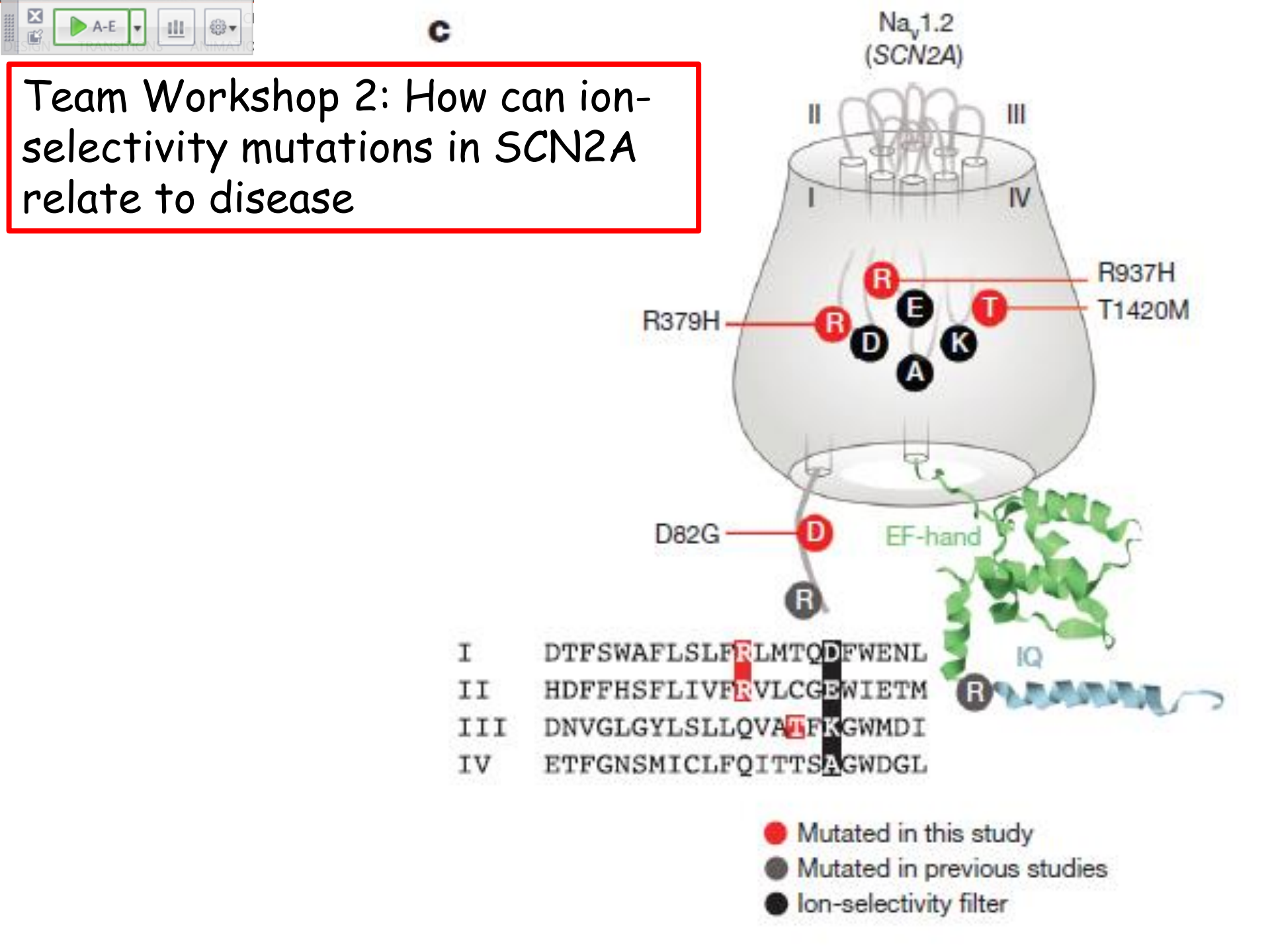
Team Workshop 1: How would you fit this data and what conclusions would you make?

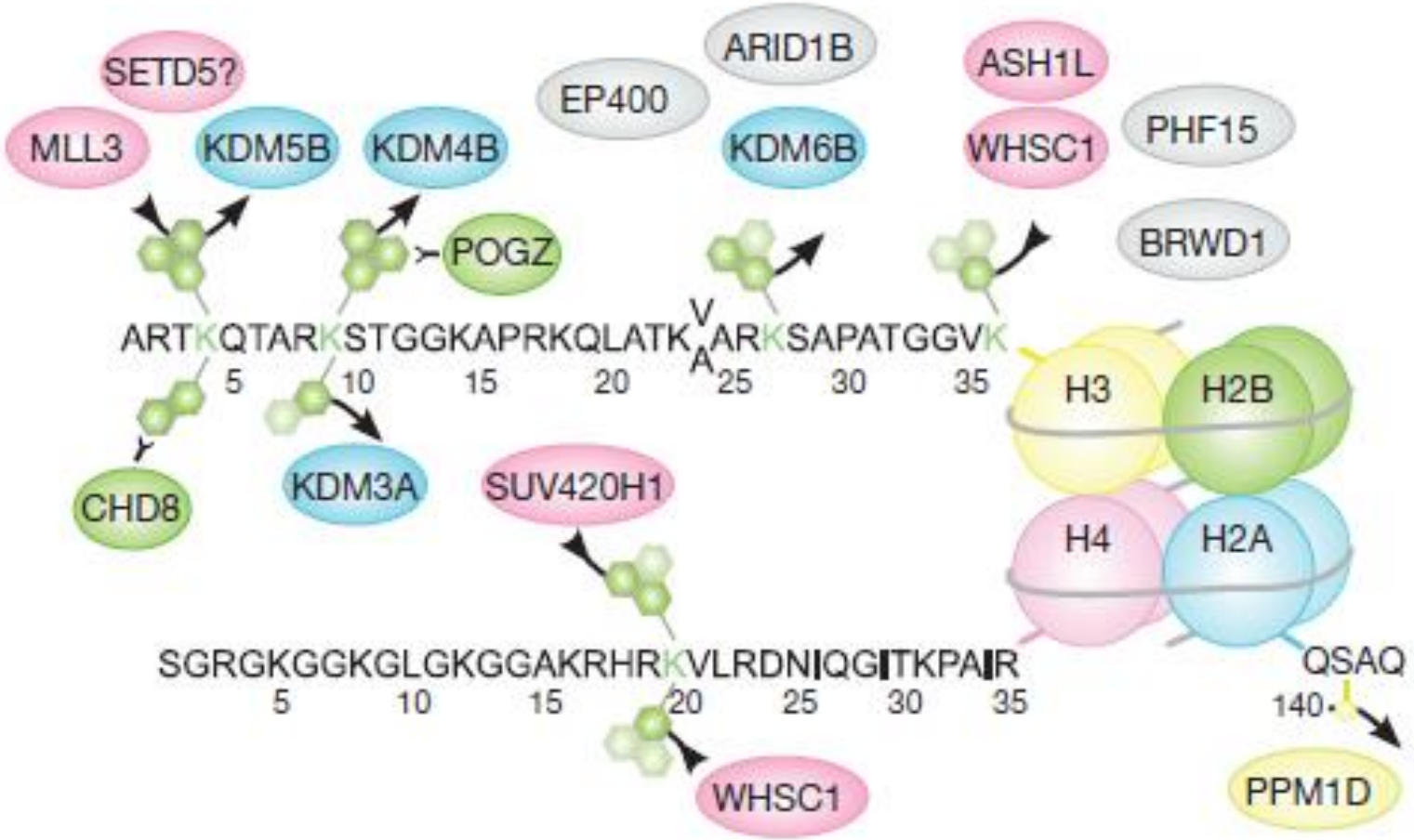


The exome sequence corresponds to

A. All the DNA of the genome except the introns
B. Only the intron sequence
C. Only the protein coding mRNA sequence
D. Only the major splice variant mRNA sequence
E. None of the above

Team Workshop 2: How can ion-selectivity mutations in SCN2A relate to disease





Team Workshop 3: List the possible outcomes of altered Lysine methylation or demethylation

Table 1 | ASD risk genes

dnLoF count	FDR ≤ 0.01	0.01 < FDR ≤ 0.05	0.05 < FDR ≤ 0.1
≥2	*ADNP, ANK2, ARID1B, CHD8, CUL3, DYRK1A, GRIN2B, KATNAL2, POGZ, SCN2A, SUV420H1, SYNGAP1, TBR1*	*ASXL3, BCL11A, CACNA2D3, MLL3*	*ASH1L*
1		*CTTNBP2, GABRB3, PTEN, RELN*	*APH1A, CD42BPB, ETFB, NAA15, MYO9B, MYT1L, NR3C2, SETD5, TRIO*
0		*MIB1*	*VIL1*

TADA analysis of LoF and damaging missense variants found to be *de novo* in ASD subjects, inherited by ASD subjects, or present in ASD subjects (versus control subjects). dnLoF, de *novo* LoF events.

Team Workshop 4: Which, if any, of these genes are sex linked?

If so why and if not, why?

Remember

- Before 12 PM of the next class day:
  - go to b.socrative.com/student/login and complete the quiz