Related article: (1 point each)
https://utswmed.org/medblog/age-matters-down-syndrome/

1. Summarize the main takeaways of this article in 1 sentence

2. This article mentions several types/causes of Down Syndrome, which of the types would not be related to germ line mutations?

3. The article points us towards potential treatments in the future to prevent this sort of accumulation of mutations in older mothers. One area of rapidly growing therapeutics is gene therapy where temporary genetic edits are made to cells with genetic defects (e.g. lung cells in cystic fibrosis patients) to cure otherwise incurable conditions. Why is this infeasible for the types of mutations mentioned here?

Paper: Parental influence on human germline de novo mutations in 1,548 trios from Iceland
Jonsson, sulem, et al., 2017
https://drive.google.com/file/d/1K6L5xvzjFaojJ7JA2JhWEyziSRAvDfCV/view?usp=sharing

Questions: (2 points each)

1. Summarize the main takeaways of this article in 1 sentence

2. APOBEC or Apolipoprotein B mRNA editing enzyme, Catalytic polypeptide-like is a family of cytidine deaminases that converts cytosine to uracil in mRNA. However these enzymes can sometimes cause mutations because the deamination of methylcytosine produces thymine in the genome. In this paper they find that APOBEC did not have a significant effect on mutational spectra. How might you expect the data to change if APOBEC activity was significant? Pay particular attention to figure 2b.

3. In figure 1e we see that paternal DNM rates are significantly higher than maternal ones. Why do you think that is? What does this tell us about how mutations tend to occur in humans? Could this possibly contribute to the differences we see as well in mutational spectra (types of mutations that occur) between mothers and fathers?

Synthesis Questions: 3 points each

1. These two articles appear to present contradictory conclusions. Can both articles be correct? Why or why not.

2. Studies similar to those conducted in ARTICLE 2., do not usually look at chromosomal aberrations. This is due to a lack of statistical power to understand them. Why could that be? What would allow the papers cited in ARTICLE 1 to look at chromosomal aberrations? How would the data they are looking at likely differ?