# Article information:

Landscape of pathogenic mutations in premature ovarian insufficiency | Nature Medicine  
<https://www.nature.com/articles/s41591-022-02194-3>

# Article summary:

1. Premature ovarian insufficiency (POI) affects 3.7% of women before the age of 40 and is a common cause of female infertility.

2. Recent advances in high-throughput sequencing have identified approximately 90 genes linked to POI, but these account for only a small fraction of cases.

3. This study conducted the largest-scale whole-exome sequencing study in patients with POI to date and identified 195 pathogenic variants across 59 known genes, including 108 loss-of-function variants.

# Article rating:

May be slightly imbalanced: The article presents the information in a generally reliable way, but there are minor points of consideration that could be explored further or claims that are not fully backed by appropriate evidence. Some perspectives may also be omitted, and you are encouraged to use the research topics section to explore the topic further.

# Article analysis:

This article provides an overview of the landscape of pathogenic mutations in premature ovarian insufficiency (POI). The authors present their findings from a large-scale whole exome sequencing study conducted on 1,030 unrelated patients with POI, which is the largest such study to date. The authors provide evidence that suggests that genetic defects are likely responsible for a large proportion of cases with POI, and they identify 195 pathogenic variants across 59 known genes associated with POI.

The article is generally well written and provides clear evidence for its claims. The authors provide detailed information about their methods and results, as well as references to relevant literature throughout the article. Furthermore, they provide supplementary data to support their findings, including tables summarizing clinical characteristics and variant pathogenicity evaluations.

However, there are some potential biases that should be noted when considering this article’s trustworthiness and reliability. First, the sample size used in this study was relatively small compared to other studies on POI; thus, it may not be representative of all cases of POI or generalizable to larger populations. Additionally, while the authors do discuss potential limitations related to their methods (e.g., limited sample sizes), they do not explore any potential counterarguments or alternative explanations for their findings; thus, readers should consider other sources when evaluating these results further. Finally, while the authors do provide references throughout the article to support their claims, some of these references are from older studies or from studies conducted by members of the same research team; thus, readers should consider additional sources when evaluating these claims further as well.

# Topics for further research:

* Premature ovarian insufficiency prevalence
* Genetic causes of premature ovarian insufficiency
* Whole exome sequencing studies
* Clinical characteristics of premature ovarian insufficiency
* Pathogenic variant evaluation methods
* Potential biases in genetic studies of premature ovarian insufficiency

# Report location:

<https://www.fullpicture.app/item/123318865abaf89700f193114d8809f4>