Diagnostic Odyssey for Rare Diseases
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BACKGROUND

- Diagnostic Odyssey is the time taken from which symptoms appear to the time taken when an accurate diagnosis is made.
- It is very common in rare diseases because the average diagnosis odyssey of the rare disease is of 5 years.
- In the US, 30 million people have rare diseases
- Due to the lack of information about rare diseases, it becomes hard to make an accurate diagnosis that delays the treatment required and leads to the death of the patient.
- Education and awareness about rare diseases are required among healthcare professionals.

Undiagnosed Diseases Program, UDP

- Started in 2008 by the National Institute of Health, NIH
- Aim to educate healthcare professionals about the signs and symptoms of rare diseases, the causes, diagnosis, and treatment options if possible.
- UDP only provides the recommendation not the treatment for rare diseases.
- High-density single-nucleotide polymorphism array, SNP, and whole genome sequencing are used by UDP to discover the diagnosis of rare diseases.
- 12 clinical sites in the US: Texas, Pennsylvania, North Carolina, Massachusetts, Minnesota, Maryland, California, Florida, Utah, Washington, Tennessee, Missouri.

UDP in different countries

- **Korea**- Korean Undiagnosed Rare Disease Program, KUDP
    - One-year program
  - started in 2017
    - at Seoul National University Children's hospital
    - 22% got confirmed diagnosed
- **India** - Indian Undiagnosed Rare Disease Program, I-UDP
    - started in 2021
  - funded by the Indian Council of Medical Research
    - 3 sites- New Delhi, Lucknow, and Hyderabad
  - approximately, 25-50% population got a confirmed diagnosis

SOLUTION

- Genome sequencing
- Genetic counseling
- Accessibility to genome testing

Cost per Human Genome

https://www.genome.gov/about-genomics/fact-sheets/D...