

Rare Disease Diagnosis

Diagnosis of Rare Diseases

Learn about the core concepts of **Rare Disease Diagnosis** and the application of medical data science.

Diagnosis Pipeline



Key Elements



Challenges

- Data Scarcity (Rarity)
- Complex Symptom Patterns
- Long Diagnosis Period
- Lack of Specialists



AI Solutions

- Few-Shot Learning Application
- Similar Case Retrieval
- Rapid Early Diagnosis
- Knowledge-Based Augmentation

Rare diseases are very difficult to diagnose due to **small patient populations and diverse symptoms**. On average, it takes **5-7 years** to reach an accurate diagnosis, during which patients cannot receive appropriate treatment.

Recent **advances in AI technology** have made it possible to address these challenges. Few-Shot Learning enables learning with limited data, and similar case retrieval allows utilization of past diagnostic cases. Additionally, integration with medical knowledge graphs has

enabled more accurate diagnoses.