# Diagnostic Odyssey for Rare Diseases

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## BACKGROUND-

Diagnostic Odyssey is the time from which symptoms appear to the time we get an accurate diagnosis. Wrong diagnosis is common in rare diseases which delays the journey from diagnosis to accurate treatment. Because of a lack of knowledge and information about rare or undiagnosed diseases, it becomes hard to get access to care and treatment at the required or targeted time. It becomes hard for medical professionals to name the diagnosis and start the initial therapy or even find the root cause or risk factors of that disease or condition. In that case, the wrong diagnosis becomes quite common which leads to wrong or inappropriate treatment approaches. Delayed or wrong treatment can also lead to the death of the patient. [1] To decrease the time for starting accurate treatment, physicians need to follow different diagnostic guidelines. Genetic testing is the initial diagnostic tool to suspect and catch the defect early. Genetic testing also helps to find the root cause or preventive measures for that disease or symptoms. This can help medical professionals to get a more specific and accurate treatment approach because it is specified to that patient's symptoms.[2] Genome sequencing is one of the genetic tests to diagnose genetic disorders in less time.[3] Awareness among healthcare providers about diagnostic odyssey is important because that can educate the physician to relate the impact of delayed treatment on a patient's life and give access to resources to diagnose the rare or undiagnosed disease. [4]Genetic counseling plays a significant role in decreasing the waiting period to initiate treatment. Providing virtual genetic counseling makes it accessible to all patients and saves patients' time. [5]About 25 million people (about the population of Texas) in the US are suffering from undiagnosed rare diseases.[6] The average length of time from symptom onset to an accurate diagnosis of a rare disease is 5 years.[7]