

## Rare Diseases

### Introduction:

A disease is considered rare if it affects one out of every 2,000 people in the population. Many of these rare diseases are hereditary and appear from birth, although some genetic diseases do not appear until late. Collectively, these diseases pose a heavy burden and have a significant impact on societies; therefore, rare diseases are a public health issue.

### Characteristics of rare diseases:

- Rare diseases are often chronic and progress over time to cause frequent health relapses.
- Rare diseases often disrupt patients' lives due to lack or loss of independence.
- Rare diseases cause pain and suffering to the patient and his family.
- Rare diseases have no effective treatment.
- There are between 6,000 and 8,000 rare diseases.
- 75% of rare diseases affect children.
- 30% of patients with a rare disease die before the age of five.
- 80% of rare diseases are hereditary causes.
- Rare diseases are the result of bacterial or viral infections, allergies, and environmental causes.

### Examples of rare diseases:

There are many types of rare diseases including mental illnesses, cardiovascular diseases, chromosomal diseases, skin diseases, infections, endocrines, urinary system diseases, bone diseases, some rare diseases such as hemophilia and albino have known names to the public, while others are named after the doctor who discovered them or even attributed to the first patient or hospital where the disease was detected, for example Harbor Syndrome.

### Number of rare diseases that can be treated:

Many rare diseases have no cure. And many others have not yet been studied by medical research. People with rare diseases often suffer from the lack of necessary and appropriate health care to find solutions to their health problems, such as the inability to accurately diagnose or the lack of appropriate medications to treat their conditions in health institutions.

### Health problems suffered by people with rare diseases:

- Difficulty in obtaining an accurate and correct diagnosis of the condition.
- Delayed diagnosis of rare diseases.
- Difficult access to medical and scientific information on rare diseases.
- Limited treatment options currently available to treat these diseases.
- Difficulty in obtaining a doctor or treatment center with expertise in the treatment of rare diseases in particular.
- Treatment is expensive compared to the cost of treating common diseases.
- Difficulty in obtaining medical, social, financial assistance or services in general, because the general practitioner who sees patients is not sufficiently aware of these diseases.
- The patient and their family may suffer socially, which adversely affects them.

### Important suggestions for patient support and health care development:

- Implementing comprehensive programs for rare diseases.
- Developing proper public health issues.
- Increasing international cooperation in scientific research.
- Acquiring and sharing scientific knowledge on all rare diseases, not only the wider spread ones.
- Developing new diagnostic and treatment procedures
- Raising public awareness of rare diseases.

- Facilitating communication among patient groups to exchange expertise and effective practices.
- Supporting most isolated patients and their parents to provide the patient with new communities
- Providing information to the community on the overall quality of the rare disease.
- Enacting legislation in favor of rare diseases such as supporting medical research and providing financial support to the patient and their family
- Providing incentives for companies to develop treatments for rare diseases.
- Collaboration and cooperation is an important and effective part in the field of supporting rare disease patients, which would be in various levels, including:
  - Collaboration on patients' level
  - Collaboration on disease level
  - Collaboration on the level of healthcare providers, specialists and patients.
  - Collaboration on the level of researchers, pharmaceutical companies and decision makers.
  - Collaboration on countries' level

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