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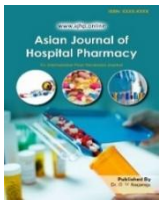

ORPHAN DISEASES: BRIDGING THE GLOBAL GAPS IN RARE DISEASE CARE ANDFUTURE PRIORITIES

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Abstract

Orphan diseases are also referred as rare disease and these are medical conditions that affect a small portion of the population. They collectively affect millions of individuals among worldwide. Orphan disease is chronic, genetic, and life-threatening, often demonstration early in life. Typically, fewer than 200,000 peoples in the United States, because of the limited count of patients, there is lack of financial support for pharmaceutical companies to invest in treatment development, and that leads in minimal therapeutic options and research, as a result, these orphan diseases are continuously unconsidered in drug development efforts. Induces to the supportive policies like the orphan drugs Act introduction. Examples of orphan disease types; genetic disorders, autoimmune diseases, rare cancer, metabolic disorders, infection disease. Characteristics of orphan disease which includes low prevalence, high mortality and morbidity, complex and often unexplained and need for orphan drug development. This article delivers an overview of orphan disease, examples, treatment, impact on patients and families, challenges in treatment and diagnosis, future direction.

***Keywords*:** Orphan disease, cystic fibrosis, Huntington disease, myasthenia gravis, neuromyelitis optica.

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Introduction

Rare disease: A small percentage of the population effects that is called rare disease. In the united state, a disease is thought rare if it affects fewer than 200,000 populations. The description focusses primarily on the number of people affected by the disease.

Orphan Disease

A rare disease that has been Orphaned by the pharmaceutical industry, means there is fine to no investment in development and research because of the small patient population and insufficiency of financial incentives.

Orphan drugs

Orphan drugs is a pharmaceutical agent that are used to treat orphan disease or rare disease. The US Orphan Drug Act (1984) knows an orphan drug as one with efficacy treating a condition that affects fewer than 200,000 people inthe United States [1-3].

Global perspectives and challenges

Initiatives to persuade pharmaceutical corporations to engage in the development of orphan medications have surfaced within the last 20 years. An important step in this regard was the Orphan Drug Act, which was first put into effect in the USA in 1983 and was later modelled by similar laws in Japan, Australia, and the European Community. Tax credits, research assistance, expedited marketing authorisation processes, and prolonged market exclusivity are among tactics used to promote the development of orphan drugs. However, in Europe, the focus on market exclusivity raises concerns about the need for additional incentives, particularly to ensure cost-effectiveness. If we define an orphan drug as one used to treat a rare disease, a rare disease could be characterized as one for which treatment is not cost-effective, or the cost exceeds £30,000 per Quality-Adjusted Life Year (QALY). This approach is crucial to avoid compromising our capacity to effectively address other diseases. Balancing the economic viability of orphan drug development with the broader healthcare landscape is essential for maintaining a comprehensive and sustainable approach to disease management [4-6].

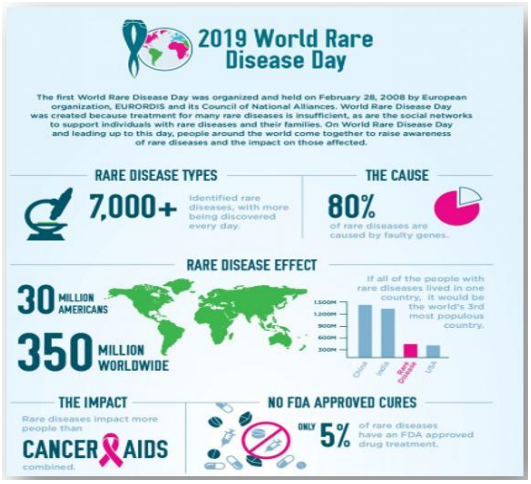

Figure 01: World rare disease day

Some examples of orphan diseases

Genetic orphan disease

Cystic fibrosis: Cystic fibrosis (CF) is a sever genetic disease that leads the body to produce thick, sticky mucus.

Symptoms: Gastrointestinal issues, recurrent respiration infections, thick mucus production.

Causes: mutation in the cystic fibrosis transmembrane conductances regulators (CFTR) gene. That causes the defective chloride transport.

Treatment

Generic drug Name: Ivacaftor

Brand name : Kalydeco

Huntington's disease

Huntington's also called as a Huntington's chorea, is an untreatable degenerative disease that is mostly hereditary.

Symptoms: depression, anxiety motor dysfunction, swelling, problem with speech and eating, memory loss.

Causes: expansion of cytosine adenine guanine, repeats in the huntington's gene, that cause toxic protein aggregation.

Treatment:

Generic drug name : Deutetrabenazine

Brand name : Austedo

Autoimmune diseases

Myasthenia gravis: Myasthenia gravis is an autoimmune condition that disrupts communicate on between nerves and muscles, leading to overall muscle weakness. In some cases, it can affect the muscles used for breathing and trigger a life-threatening situation known as a myasthenic crisis.

Symptoms: Myasthenia gravis mainly causes muscle weakness that comes and goes, especially as the day goes on. It often starts with eye-related issues like drooping eyelids or blurred/double vision. Muscles used for facial expressions, talking, and swallowing can also be affected. In some people, the weakness may spread to the arms, legs, or even the muscles used for breathing, which can lead to a serious condition called a myasthenic crisis.

Causes: Myasthenia gravis happens when communication between the nerves and muscles breaks down. It's an autoimmune disorder, meaning the body's defense system wrongly targets and attacks its own healthy tissues, disrupting normal muscle function.

Treatment

Generic drug name: Pyridostigmine

Brand name : Mestinon

Neuromyelitis optica

Neuromyelitisoptica (NMO), also known as Devic's disease or neuromyelitis optica spectrum disorder (NMOSD), is a condition that affects the central nervous system. It causes inflammation, mainly in the optic nerves - which link the eyes to the brain - and in thespinal cord. This disorder happens when the immune system mistakenly attacks healthy cells, particularly in these areas, and can sometimes also affect parts of the brain.

*Symptoms:*Optic neuritis causes symptoms due to swelling in one or both of the optic nerves.Myelitis leads to symptoms as a result of inflammation in the spinal cord.When NMO impacts parts of the brain like the hypothalamus or brainstem-areas that manage essential automatic body functions-it can lead to problems with brain activity.

*Causes:*It happens when your immune system attacks nerves in your eyes or your spinal cord. It's not known what causes it. Sometimes it happens after a virus such as flu. NMO is not an inherited condition.

Treatment:

Generic drug name: Eculizumab

Brand name :Soliris [7-10].

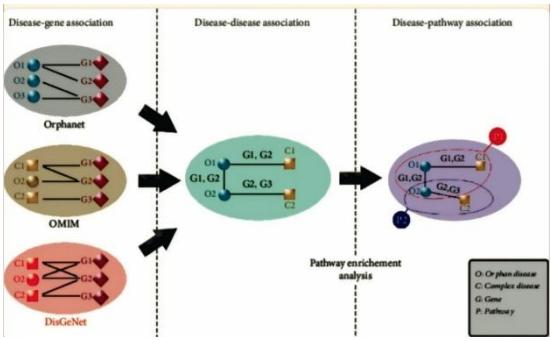

Figure 02: A computational and integrative method was used to explore connections between diseases and genes, relationships among different diseases, and how diseases are linked to biological pathways

Impact on patient and families

Acrosssectional study in the valencian region,spain. Families with orphan disease have unmet requirements that are often overlooked by the medical professionals.A total number of 163 survey were acquired from patients visiting primary care centre in the valencian region (spain), in period of 2015-2017, with a conformed or suspicious diagnosis of orphan disease of the 84.7% with a accommodate diagnosis, 50.4% had a diagnostic late exceptional one year , and it was more frequent among adults (62.2%),families with young patients were in a poor

economic situation, with less incomes and higher rate of disease - related price (300on on average).

Impact on patient

Physics health:Many orphans disease is long term, progressive, decadences, and potentially life-threating, causes to a loss of autonomy and effective quality of life.

Social life: Orphan disease can be causing social isolation, prohibit from community activity, and struggling in maintaining relationships.

Psychological well-being: Living with an orphan disease can lead importances emotional distress, contain depression, anxiety and isolation.

Impact on families

- Emotional burden: Families of individuals with rare diseases often go through intense emotional stress, feeling anxious about understanding the condition, what the future holds, and the limited treatment options available.
- Financial strain: Dealing with rare diseases can be very expensive, and the high costs often create serious financial pressure on families, sometimes affecting their ability to meet basic needs.
- Impact on siblings: Brothers and sisters of those with rare diseases can face emotional challenges, feel socially isolated, and may have their schooling or career plans interrupted.
- Caregiver burden: Looking after a child or family member with a rare disease can be exhausting both physically and emotionally, often resulting in stress and burnout for the caregiver.
- Family dynamics: Rare diseases can change how a family functions, often needing changes in daily schedules, how the home is managed, and who takes on caregiving roles.

In short, orphan diseases don't just affect the person diagnosed-they also place major strain on the whole family, bringing difficulties to many parts of their daily lives [11-14].

Challenges in treatment and diagnosis

Orphan diseases give unparalleled challenges in both treatment and diagnosis because of their complex nature and rarity.

Diagnosis Challenges

- ***Limited awareness***: Medical practitioners may not be familiar with specific orphan diseases. That cause to the underdiagnosis or belated diagnosis.
- ***Specialized testing***: Diagnosis frequently requires specialized assessment and procedures that may not be willingly available, forward late the diagnostic process.
- ***Heterogeneity***: The comprehensive characteristics and manifestations in some orphan disease can affect diagnosis more challenging.
- Lack of research data: Finite Research data on orphan disease obstructs the development of diagnostic tools and treatment strategies.

Treatment challenges

- Clinical trial limitations: Detecting adequate patients for clinical trials can be challenging, specifically when targeted treatment options are being examine.
- Lack of specific therapies: Many rare diseases shortage specific treatment and offlabel use of current medication maybe the only option, which can be hazardous and not fully efficacious.
- Regulatory hurdles: The regulatory process for progressing and accepting new treatment for orphan disease can be complicated and prolonged.
- Patient recruitment: Recruiting member for clinical trials can be tough due to the rarity of the disease and the dissipate patient population.
- Funding and investment: Need of financial incentives for pharmaceutical companies to evolve treatment for rare diseases advance limits the availableness of new therapies.
- Accessibility: Even when treatments are provided, they may not be reachable to all patients due to geographic limitations, cost and other barriers [14-17].

Future directions

Earlier and Faster Diagnosis:

- Genomic Sequencing:New developments in genetic sequencing are making it easier and quicker to diagnose rare diseases, helping patients get earlier access to appropriate treatment.
- Data Science and AI: By using artificial intelligence and machine learning to examine large collections of genetic and medical data, researchers can now detect rare disease patterns more effectively, speeding up the diagnosis.
- Standardized Nomenclature and Coding: Creating consistent names and coding systems for rare diseases supports better classification, helping to distinguish conditions and pinpoint specific patient groups.

Accelerated Therapy Development:

- ***Personalized Treatment***: treatment. Developing therapies based on an individual's unique genetic and molecular characteristics is becoming a key focus in rare disease.
- **Support for Orphan Drugs**: Government and regulatory incentives are encouraging pharmaceutical companies to create new treatments for rare conditions, supported by scientific innovation.
- **Outcome-Based Care Models**: Shifting toward care systems that focus on treatment results helps make rare disease therapies both more effective and more accessible to patients.

Improved Patient Outcomes and Quality of Life

- Use of Digital Tools: Devices like wearables and remote monitoring systems are transforming patient tracking and data collection in clinical research and everyday care.
- Whole-Person Care: Treating rare disease goes beyond physical symptoms; addressing mental health, emotional support, and social needs is equally important.
- Patient and Family Involvement: Advocacy organizations play a vital role in shaping policies, spreading awareness, and ensuring patients and caregivers have the support and resources they need.

Enhancing Rare Disease Research

- Data Sharing and Collaboration: Creating strong systems for sharing information among researchers, clinicians, and patient groups can speed up progress.
- Global Coordination: International organizations play a vital role in facilitating global collaboration and coordination.

Overcoming Obstacles

- Streamlining Regulations: Simplifying drug approval processes and encouraging early communication between regulators and industry can help new therapies reach patients faster.
- Building Capacity: Investing in people, technology, and infrastructure is essential for sustainable rare disease research and care.
- Ethical Considerations: Researchers must address ethical concerns, particularly in pediatric clinical trials, to ensure responsible and safe practices.

Conclusion

Orphan diseases, or rare disorders, are illnesses that affect a very small segment of the population-usually fewer than 1 in 2,000 people. Though individually uncommon, over 7,000 such conditions have been identified, collectively affecting millions across the globe. These diseases often have serious effects on patients and their families, causing long-term physical suffering, emotional stress, and significant financial strain due to persistent symptoms, disabilities, and lack of specialized treatment options.A major hurdle in managing rare diseases is the difficulty in reaching a timely diagnosis, which is often delayed by limited awareness, a shortage of expert clinicians, and inadequate diagnostic tools. Treatment choices are also scarce, as only a few therapies are available, and developing new ones faces economic, scientific, and regulatory barriers. Orphan diseases can be grouped by their underlying cause-such as genetic, autoimmune, or infectious origins-or by the body systems they affect, like the nervous or blood systems. To improve outcomes for those living with rare diseases, future strategies must include more investment in research, stronger global collaboration, advancements in diagnostic methods (including genomics and artificial intelligence), and policies that encourage drug development. Ensuring patient-focused care and international data sharing will be essential to overcoming current challenges and making real progress in rare disease management.

Conflicts of interest

None

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