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RARE DISEASES AT THE EMERGENCY DEPARTMENT: TIME FOR ACTION.

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Abstract

In Europe rare diseases (RD) are defined as those that affect one in two thousand individuals or fewer. Globally, they affect around 6% of the population, and this means that, collectively, healthcare providers deal with a considerable number of patients with a rare disease, of which over 70% are genetically determined. Aim of this paper is to develop a proposal for a national flowchart applicable to patients with known or unknown rare disorders at the ED.

KEYWORDS: Rare Diseases, Chromosomal, Abnormalities, Healthcare and Patients

INTRODUCTION

In Europe rare diseases (RD) are defined as those that affect one in two thousand individuals or fewer. Globally, they affect around 6% of the population, and this means that, collectively, healthcare providers deal with a considerable number of patients with a rare disease, of which over 70% are genetically determined. A recent calculation of the cumulative prevalence at a global level based on the Orphanet epidemiology data file estimates that, regarding the 2017 population, there is a minimum prevalence of 3.5–5.9%, which corresponds to 263–446 million persons affected worldwide.

According to a National Vital Statistics Report in the United States published 2016, congenital malformations, deformations and chromosomal abnormalities accounted for 22.2% of all deaths in neonates, 20.8% of all infant deaths, 10.7% from 1 to 4 years of age. The European Conference on Rare diseases in 2005 analyzed life expectancy of 323 rare diseases and found that 25.7% of them were potentially lethal before 5 years of age, 36.8% lead to a reduced life expectancy, while 37.5% were compatible with a normal lifespan.

The annual cost of orphan drugs was estimated $15 billion in 2007, $30 billion in 2013 in the United States alone. In 2017, the worldwide orphan drug sales were estimated

at $125 (about 15.9% of all nongeneric prescribed drug sales). According to the same report, the mean cost per patient per year was 4.8 greater for orphan drugs than for nonorphan.

An estimated 15.5 million children and adults living in the United States and affected with any of the 379 rare diseases in 2019 accounted for a total economic burden of $997 billion, (direct medical cost of $449 billion (45%) $437 billion (44%), $73 billion due to non-medical costs (7%), $38 billion (4%) related to healthcare costs not covered by insurance).

They are often scarcely taken into consideration by most of physicians, especially at emergency departments, with a consequent difficulty in both recognizing and treating them. Lack of practice guidelines or consensus were considered the most important causes of diagnostic delay or missed diagnosis of rare diseases.

In a recent survey conducted among 539 physicians in China 98.5% considered their knowledge about rare diseases insufficient. Patients with rare diseases are at high risk of diagnostic errors and delays.

Physicians at the emergency department (ED) should remember that not all rare diseases constitute a diagnostic dilemma and that not all the patients with undiagnosed disease have a rare disease. Moreover, patients with RD can present with symptoms and signs that strongly mimic common diseases, while others can develop with multisystem slowly evolving symptoms and signs that make diagnosis difficult and elusive.

Particularly:

- Some rare disorders can be diagnosed for the first time at the ED due to acute appearance of symptoms and signs (for example rhabdomyolysis revealing type V Glicogen Storage Disease or Mc Ardle Disease).
- A patient with a diagnosed rare disease can be evaluated at the ED due to sudden symptoms and signs which can be partially/totally related or not related to RD.

Rare diseases management is not familiar to physicians acting in ED, and taking care of RD patients in the emergency setting can prove challenging for them. Emergency situations can result from the rare disease itself or be unrelated to it, but most often need particular measures to be taken.

Besides of emergency guidelines, the French Ministry of Health has produced personal emergency cards for a number of rare diseases during the first national plan for rare diseases. These cards include summarized recommendations in case of emergency intended to physicians, as well as relevant personal information to be completed by a practitioner in

the center of reference. It includes some recommendations for patients intended to prevent emergency situations related to the disease. Emergency cards are also available on the Orphanet website.

AIM, MATERIALS AND METHODS.

To develop a proposal for a national flowchart applicable to patients with known or unknown rare disorders at the ED.

RESULTS AND DISCUSSION.

A small emergency card explaining the main characteristics of rare disease should be prepared for all patient. This document should also contain summarized recommendations in case of emergency intended to physicians, based on the project and following the French model. It also should contain information's about life-saving drugs, and contraindications. They should also contain details about possible pharmacologic trials in which patients are currently enrolled.

- All the physicians at the ED should be educated at promptly suspect and recognize possible rare disorders and acute complications of them.
- Dedicated flowcharts should be planned and promptly activated for patients with rare disorders, especially with high physical/intellectual disability.
- The presence of caregiver should be allowed even during pandemic period for patients with rare disorders and barriers to effective communications.
- Multidisciplinary team with expertise in treating rare disorders should be alerted, when possible, in order to guarantee quick evaluation and treatment and to reduce waiting times

CONCLUSIONS.

We think that Emergency Guidelines dedicated to patients with rare disorders should be discussed and approved. Special flowcharts for patients with rare diseases, especially with high disability, should be approved in order to promptly recognize, monitoring and treat them, reducing mortality and complications.

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