Commentary

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To Avoid the Recurrence of Myeloma Multiplex, And Due To the Frequent and Serious Angioedema Attacks

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Description

C1-INH-AAE, or acquired angioedema caused by C1-inhibitor deficiency, is a rare condition that frequently manifests alongside hematological conditions. It is diagnosed with complement testing, and its treatment consists of treating the underlying disease and treating angioedema attacks. Due to an upper airway edema that did not respond to conventional treatment and necessitated a tracheotomy, the 59-year-old male patient was referred to the Hungarian Angioedema Center of Reference and Excellence. C1-INH-AAE was diagnosed at our center, and myeloma multiplex was found to be the underlying condition. For the past 15 years, he has been followed up on. His complement parameters returned to normal following the first treatment series (chemotherapy and stem cell transplantation), but five years later, they began to deteriorate and the angioedema attacks recurred. The patient received a second course of rituximab treatment in order to prevent a recurrence of myeloma multiplex and the severe and frequent angioedema attacks. He didn't have any more angioedema attacks as a result of it, and his complement values returned to normal. Angiotensin-convertase-catalyst inhibitors can set off C1-INH-AAE. The repeat of the hidden illness or the angioedema assaults can be anticipated by the standard development of the supplement boundaries. As a result, the underlying disease's treatment can begin before severe symptoms appear. Nearly 400 million people worldwide suffer from rare diseases, which have devastating effects on patients and their families.

Arteriovenous Malformations

The research community frequently overlooks these diseases, despite their collective commonality. As an example of how rare disease research should be conducted, we present the PACS1 Syndrome Research Foundation's ongoing endeavors. Due to high costs and clinical uncertainty, access to medicines for rare diseases—also known as "orphan medicines"—has proven challenging. During Marketing Authorization (MA) and Health Technology Assessment (HTA), some healthcare systems are implementing specialized pathways and/or processes to maximize market access to these medicines. This study aims to determine whether the presence of specialized pathways and processes at the MA and HTA levels is associated with more favorable funding recommendations and a quicker time to market access by contrasting one setting in which these medicines are considered to be "orphan" (Scotland) and another in which they are considered to be "non-orphan" (Canada). The data were extracted from publicly accessible sources, and a matched sample of 116 medicine-indication pairs with MA approval from 2001 to 2019 in Europe and Canada was found. For data analysis, descriptive statistics were utilized. Except for one instance in Scotland, all medicines were commercially marketed in both countries. While Scotland had a higher percentage of orphan medicines with favorable HTA recommendations (68.1%) than Canada (60.4%), Canada had a higher percentage of negative HTA recommendations (20.7%) than Scotland (15.5%). Between settings, there was little agreement on HTA recommendations and the main reasons for them.

There was a lower rate of negative HTA recommendations for medicines with specialized MA approval than for medicines with standard MA approval in both countries. Canada had a faster time to market access than Scotland, but both countries had slower timelines for medicines with specialized MA approval than for medicines with standard MA approval. Without taking into account other aspects of the healthcare system and variations in the decision-making processes between settings, it is, however, unclear whether the presence of orphan designation and HTA specialized processes alone could result in favorable funding recommendations. To maximize access to orphan medicines, comprehensive strategies and improved alignment of evidentiary requirements across regulators are required. The rare vascular multisystemic disorder known as Hereditary Haemorrhagic Telangiectasia (HHT) causes epistaxis, blood loss-related anemia, and arteriovenous malformations in the lungs, liver, and brain. The European Reference Network for Rare Multisystemic Vascular Diseases serves patients with HHT, whose prevalence is estimated to be 1/6000, or approximately 85,000 Europeans. HHT treatments span a wide range of medical, surgical, and interventional specialties and are based on clinical manifestations. In severe cases, in addition to local treatments in the nose, intravenous bevacizumab has been suggested as a treatment option. The objective of this article is

to evaluate the use of intravenous bevacizumab in patients with HHT in 2022 based on the data that are currently available. In a variety of medical image analysis tasks, deep convolutional neural networks (ConvNets) have demonstrated performance that is at the cutting edge. A lot of labeled data are partly to blame for the success. However, it is difficult to obtain a large amount of labeled data from some medical images, such as a rare disease case of actinic keratosis, in hospitals.

Rare Diseases

As a result, the method for training a network to classify medical images with very little data—three or five samples per class—is crucial but receives little attention at the moment. A difficulty-aware Meta learning approach for the classification of rare diseases using dermoscopy images is the subject of this book chapter. The fundamental idea is to train a model on a variety of learning tasks so that it can solve new problems, such as rare disease classification, with just a small number of labeled samples. We introduce the Difficulty-Aware Meta Learning optimization loss (DAML), which is inspired by recent advancements in Meta learning. The observations that the contribution of various task samples is varied serve as the inspiration for our novel difficulty-aware metaoptimization loss. Our method is able to quickly focus on the difficult tasks and downweight the well-learned ones by dynamically monitoring the scaling factor. The ISIC 2018 skin lesion classification dataset is used to evaluate our method. With only five samples per class, the model can quickly adjust to classify unseen classes with a high AUC of 83.3%. We also provide examples of how our method can be utilized in actual clinical practice for the classification of rare diseases. An increasing number of people suffer from rare diseases. Reaching experts and organizations with expertise in a particular disease is a major issue for patients and their caregivers. Consequently, caregivers, who are frequently members of the patient's family, gain a wealth of knowledge about the disease through personal experience. Caregiver-Matcher is a proof of concept that offers a smart solution for constructing a network of caregivers connected by a graph neural network-based matching mechanism. The parental figures and their involvement in uncommon illnesses are portrayed by hub highlights. The platform invites associations and care facilities to share their knowledge.

Pain may progress from acute to chronic in patients with rare musculoskeletal or neuromuscular diseases; the latter poses additional difficulties for both patients and medical professionals. We looked at how pain is currently understood in relation to ten rare, non-cancerous, infectious disorders; Ehlers-Danlos Syndrome, Osteogenesis Imperfecta, Achondroplasia, Fibrodysplasia Ossificans Progressiva, Fibrous Dysplasia/ McCune-Albright Syndrome, Complex Regional Pain Syndrome, Duchenne Muscular Dystrophy, Infantile- and Late-Onset Pompe disease, Charcot-Marie-Tooth Disease, and Amyotrophic Lateral Sclerosis are some of the conditions that fall under this category. We described pathologic and genetic factors, pain sources, phenotypes, and, lastly, existing therapeutic approaches by integrating natural history, cross-sectional, retrospective, clinical trials, and case studies. Additionally, we described pain phenotypes. Even though rare diseases have distinct core pathologic features, there are a number of common pain phenotypes and mechanisms that can be prospectively investigated and therapeutically targeted simultaneously. The evolving nature of pain phenotypes in rare musculoskeletal or neuromuscular illnesses is also explained, as are clinical and research strategies that may make it easier to accurately diagnose, monitor, and treat pain.