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Training for a Neurology Career in a Rare Disease: The Role of "Cyberconsults"

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Background

One little known fact about rare diseases is how numerous they are: more than 6,800 conditions correspond to the definition of the Orphan Drug Act enacted by the US Congress in 1983. A disease is considered rare –or orphan- if it affects fewer than 200,000 Americans[1] or fewer than 1/2000 people in the European Union.[2] According to the national organization for rare disorders (NORD), it is estimated that between 6–8% of the population is affected by a rare disease, which amounts approximately to 25 million people in the US.[3]

Developing clinical expertise in a rare disease

Who are the physicians taking care of these patients? Specific interest in a rare disease may arise as a particular opportunity to resolve unanswered questions, or may be born from the experience of having a friend or family member with such a condition. However, there are many obstacles to overcome before this interest can become part of a successful medical practice. First, one has to become an expert in the subject. This is difficult since the disease is -by definition- infrequent, and only a few cases may occur every year in any geographic area. Getting referrals of such patients requires having a larger practice in a subspecialty to which this disease belongs, but this may not be enough. For example, a movement disorder specialist interested in Huntington's disease will have to "compete" for those patients with other subspecialists in the vicinity. To build up a large contingent of rare disease patients, the subspecialist may have to offer additional benefits to those patients and their referring physicians, such as an opportunity to enroll in research studies or clinical trials, or participation in a patient support group.

Obtaining research funding for a rare disease

A second obstacle to succeeding in these endeavors is to obtain funding for this research. Most rare diseases do not have a dedicated society or association, and research grant

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CONFLICT OF INTEREST:
None

applications to government agencies like the National Institutes of Health (NIH) or the National Science Foundation (NSF) will be considered by study sections often devoid of members with specific expertise on the topic and will compete against proposals aiming to study common conditions affecting millions of people. Nevertheless, grant applications may be considered favorably if they use the rare disease as a model to understand pathways or mechanisms that may inform universal principles applicable to common illnesses.

Achieving academic prominence in a rare disease

Patient referrals in rare diseases practice

Over time, the newly recognized experts will receive referrals of patients from a wider network of physicians, leading to over-representation of the rare disease in their clinical practice. Patients and their families will also reach out to experts directly after diagnosis, seeking confirmation or a second opinion. This often occurs through support group websites or through government-sponsored organizations like NORD[3] or EURORDIS[2]. Experts are often called upon to publish descriptions of these diseases in lay language or to talk with the lay press, providing patients with another opening to contact them directly. There are various internet-based communities which help provide support for patients with rare diseases, but also create opportunities for researchers. Finally, social media may play an increasingly large role in the dissemination of information and in communications from patients and referring providers. Once experts have accrued a solid patient base in a rare disease, they are ready to train their first fellow.

Training fellows in a rare disease

About 78% of neurology residents in the US enter fellowships[4]. Residents seldom have an interest in a single disease; instead, they are attracted to a particular sub-specialty. Most ACGME (Accrediting Council for Graduate Medical Education) or UCNS (United Council for Neurological Specialties) accredited fellowships are designed to provide comprehensive training in the field of subspecialty. Within those programs, fellows may wish to take advantage of mentors who have taken on a rare disease as their focus of clinical and research activities. The fellows can learn about the differential diagnosis and management of the rare disease in their mentor’s clinic and may contribute to a research effort in the laboratory. In other cases, physician-scientists may want to do a research fellowship specifically on that rare disease. They will be on the front lines of helping patients with this disease, for which their mentor is contacted from around the world for advice on cases via email or telephone. We will use our own experience with “cyberconsults” to discuss how such opportunities can help train neurology specialists in rare diseases and how they inform us about the ethical and legal considerations surrounding the use of such remote-access technology in patient care.

An example of rare disease “cyberconsults”: Progressive multifocal leukoencephalopathy

In our neuro-HIV and neuro-infectious diseases fellowship, we have a special focus on neurologic diseases caused by the polyomavirus JC Virus, including progressive multifocal

leukoencephalopathy (PML), JCV granule cell neuronopathy (JCV GCN), JCV encephalopathy (JCVE) and JCV meningitis (JCVM).[5] PML is considered an orphan disease, occurring in approximately 4000 people per year in the US and Europe combined; JCV GCN, JCVE and JCVM have only been reported in small numbers of patients, and there are no proven treatments for these conditions.

Over the past academic year (July 2013 to June 2014), we received 43 requests for remote consultation, including email or phone contacts from a patient, family member, or physician outside our institution. Eight patients (19%) were from within the state. Twenty-five (58%) were from within the US but outside of Massachusetts. Ten (23%) were from other countries including Canada, Australia, Finland, Italy, Germany, United Kingdom, Hungary and Thailand.

Of the 43 patients, 33 (77%) had one of the JCV syndromes (PML, JCV GCN, JCVE, JCVM), 8 had other diseases and 2 had insufficient data to comment on the diagnosis. A total of 6 patients were subsequently seen in our clinic for a consultation, 5 of whom had a JCV syndrome. Eleven of the 33 patients with JCV syndromes were enrolled in our research studies and we received autopsy specimens on 3 such patients.

Interestingly, 13 consults (30%) were for atypical presentations, while the majority were for second opinion and inquiry regarding any clinical treatment trials. When we were contacted by physicians, they were most often neurologists, infectious disease specialists or hematologist-oncologists.

Legal and practical issues of “cyberconsults”

There are several important considerations for physicians who perform remote consultations (defined as those for which there is no face-to-face contact with the patient). Patients and families are often devastated by the diagnosis of a rare disease for which there is no cure, especially if they live in areas devoid of expertise in this field. Therefore, their calls often contain much anguish as well as expectations. The first limitation that needs to be clearly understood is that it is not possible to practice medicine over the phone or the internet if the patient has not been evaluated in person by the expert. Therefore, the only advice we can render is to describe the steps we would take to establish the diagnosis or determine management, if we were to see a similar presentation in our clinic. Another concern for such email or telephonic consultation is malpractice liability. The disease expert as well as the referring physician should both realize this limitation and decisions for medical care should be made in light of the fact that the expert has not seen the patient and may not have access to all of the medical records. The expert should request as much primary data as possible and clearly state the limitations of the advice. When patients or their families contact an expert directly, various practical issues arise, one of which is the concern about providing an opinion, which will likely be regarded as very valuable by the family, without a full assessment. We avoid that by asking to communicate directly with the single physician who is most involved in the patient’s care.

Limitations and rewards of “cyberconsults” for rare diseases

There is no direct financial compensation for such consults in most cases. A significant amount of time may be spent trying to gather relevant data, if a physician is not involved in the communication from the other end. The process may be dissatisfying, if one does not get appropriate information to render any advice, or if there is not follow-up on the patients. Several experts may be contacted simultaneously about the same case, and formulate different opinions based on the information they have been gathering separately. This may create more confusion for the patient and family.

The rewards in these cases are most often non-financial. First, this kind of work leads to the deep satisfaction of helping patients worldwide who do not have access to similar expertise locally. Second, it provides the expert with an expanded pool of patients from which to recruit for research and treatment studies. Third, interesting presentations and novel clinical syndromes can be found through such consults, leading to new avenues of research and publications.[6, 7] Fourth, grateful patients and their families may choose to support through philanthropy the clinical, research and training programs from which they benefited. Fifth, such consults also help widen one’s professional network, and as has been our experience, the referring physicians are likely to send additional patients. Sixth, by establishing a world-wide reputation in a field, the physician is likely to receive a larger share of referrals for cases that occur locally, and may bring in some patients who can travel longer distances. This enriches the rare disease clinic and fellowship and provides a larger platform for research efforts. Finally, these consults are of significant value for trainees in these rare diseases. As the fellows trained in a rare disease move to other institutions and become the local experts, it is a good opportunity to expand their professional horizon as they interact with physicians of different specialties throughout the country and around the world.

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