A DIFFERENT VIEW

The paediatrician’s role in support groups for rare diseases
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MAGNITUDE OF THE PROBLEM
Rare diseases have come to be called orphan diseases because pharmaceutical companies are reluctant to ‘adopt’ treatments for them because the financial return does not justify the investment. Orphan diseases have been defined in the United States as individual conditions with a prevalence below 7.5 in 10 000 Americans (1). In the European Union, orphan diseases are defined as life-threatening or very serious diseases with a prevalence below 5 in 10 000 Europeans (2). Examples include cystic fibrosis, fragile X syndrome, neurofibromatosis type 1, sickle cell disease, spinal muscular atrophy, Prader-Willi syndrome, retinoblastoma, Rett syndrome, tuberous sclerosis and Williams syndrome, to name but a few.

A special category of ‘ultra-orphan’ diseases has been suggested by the United Kingdom National Institute for Clinical Excellence for diseases with a prevalence of below 1 in 50 000. It includes lysosomal storage diseases, for which research has yielded promising developments. One source estimates that there are currently about 7000 orphan diseases (www.orpha.net).

Despite the low prevalence of each of them, orphan diseases are collectively estimated to affect about 30 million Europeans and 25 million North Americans (3). Most are severely disabling with a chronic course, and many are degenerative and life-threatening. Manifestations are varied, often affecting several domains in a single patient, e.g. communication, learning, nutrition, sleep, mobility, pain, etc. They have a significant impact on patients, their families, the community and health services (4).

SUPPORT AND ADVOCACY GROUPS
The diagnosis of rare diseases is often difficult and delayed (5). Management is typically complex, involving multidisciplinary programmes, and there is currently no curative treatment for most of these diseases. Patients and their caregivers often report that they experience obstacles in receiving adequate medical and social care, despite relative satisfaction with health professionals’ kindness and readiness to help (6). As a result, patients’ organizations that focus on rare diseases (individually or in general) have been set up in most developed countries to provide emotional support and guidance, improve quality of life and health outcomes as well as spread information, contribute to clinical research, establish patient registries, promote research funding and development of specific treatments and influence social and health policy.

These groups have thus become an important healthcare resource. For patients with rare diseases, some support groups are better sources of information than physicians (7). They champion self-education and self-responsibility, foster patients’ assertiveness and initiative, and enable members to assist others. Some professionals appear to view patients’ and families’ access to these resources as challenging their medical role as bearers of valid practice and counselling. Other professionals might ignore them altogether. We, on the other hand, see potential benefits in better understanding the dynamics of collaboration between support groups and paediatricians to the benefit of patients with rare diseases.

In Europe, currently more than 1600 such organizations are dedicated to one or more rare diseases, federated by the European Organisation for Rare Diseases (Eurordis, www.eurordis.org) and various national associations, and in the United States more than 1200 organizations linked to major networks, such as the National Organization for Rare Disorders (NORD, www.rarediseases.org) or the Genetic Alliance (www.geneticalliance.org). The justification for specific groups for families affected by rare diseases seems to be the feeling of social exclusion experienced by many of them, even when interacting with those concerned by other disorders. In particular, their children’s special...
needs appear to be poorly known or recognized, including by schools and institutions, social services and policy makers.

The success of these groups, catalyzed in the last few years by media exposure, relies on the value of emotional support from individuals who share the same experiences. These groups provide their members (peers with a common experiential bond) with a source of support, as well as an opportunity to improve the quality of life with the disease. These groups can lead to changes in attitudes towards health-related problems, promote public awareness and may develop guidelines for health policy making. The groups’ activities are commonly centered on gathering and disseminating information on the disease and social and medical initiatives for managing related problems facilities, as well as optimizing primary care and use of health service. A distinctive feature is organization based on equality between group members in sharing the problems, contrasting with the polarity between the helper and the persons being helped that characterizes the conventional relationship between the professional and the patient’s family. This may help increase autonomy of patients and families in their search for appropriate management of everyday life problems and optimization of future prospects.

**PAEDIATRICIAN’S ROLE**

The autonomy gained by patients and families through support and advocacy groups has been described as empowerment by Aymé et al. (8), who recently suggested that patients with rare diseases and their support organizations are among the most empowered groups in the health sector, mainly as a result of their own fight for recognition and improved care. This process does not imply independence from medical professionals. Rather, it may contribute to redefining their partnership. This is consistent with the notion of the expert patient (9), which needs to be reconciled with professional understanding of disease. Even without any direct reference to disease, the American Academy of Pediatrics (10) has recommended that paediatricians engage in a relationship with parents based on collaboration and shared decision making so that they feel and become more competent. The Academy also urges paediatricians to participate in community-based family support programmes. Within these programmes, paediatricians should provide technical advice on health and safety aspects of services, serve as a source of professional information for families and learn from these programmes how to best contribute to the healthy development of children, families and communities (10).

Collaboration with support and advocacy groups is based on mutual confidence and respect. It is critical that paediatricians avoid confusion of roles and not act as patients’ physicians. It requires from them readiness to be humbly receptive. Paediatricians should be prepared to question previous notions and consider their collaboration as potentially useful (or even necessary) for the quality of their work.

In this context, they have the opportunity to develop tools that allow caregivers to better understand professional jargon and relevant medical and scientific notions. They participate in conferences relating to specific themes and questions, e.g. communication, feeding, epilepsy, orthopaedic issues, schooling issues, leisure, transitions or projections into the future. They help disseminate information, including at the clinical and academic levels. They help create or reinforce networks linking parents and professionals (e.g. in institutions) in order to promote exchange of opinions and good practice. They advocate the need for clinical as well as basic research. If it is relevant to set up a scientific board, they provide scientific answers to health-related questions put forward by members in this context and may evaluate research funding applications. They advocate patients’ and families’ rights, including at the political level. Confrontation, discussion, exchange and common participation in projects with parents are likely to change their attitude towards patients, caregivers and disease. Support and advocacy groups represent a model of informal education. Collaborating with them is likely to contribute to the paediatrician’s own continuing education, particularly in communication skills and perception of issues (emphasizing a family-oriented perspective).

**CONCLUSION**

Improving the outcome of rare diseases is of the utmost concern to paediatricians and patients’ families. Their collaboration within support and advocacy groups can enhance progress to this end if paediatricians accept the goals of improving knowledge through research, developing appropriate information for patients, health professionals and the general public, and promoting access to screening and diagnostic testing as well as to quality treatment and social benefits.

**References**

1. Food and Drug Administration. Available at: www.fda.gov/orphan/oda.htm