SPECIAL COMMUNICATION

Zebras on the Commons: Rare Conditions in Family Practice

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Background: **Family physicians (FPs) specialize in the management of common problems, but we know little about their role in the care of patients with rare conditions.**

Objective: **To describe the roles FPs play in the identification and management of patients with rare conditions in a typical practice.**

Methods: **Office record review of 100 patients with rare conditions in the everyday, community-based, private practice of 4 FPs. Analysis of patient demographic characteristics, diagnoses, and the roles played by the FP in the patient's care, including diagnosis, treatment, referral, and long-term patient management.**

Results: **These FPs cared for patients with a wide variety of rare disorders across the spectrum of patient age and sex, organ system involved, and medical specialty area. FPs identified the problem in 89%, diagnosed the disorder in 54%, provided acute care in 56%, and provided continuing care for 76% of patients. FPs consulted other physicians in 85% of cases. The condition was life threatening in 58% of patients.**

Conclusions: **Family physicians provide a broad range of services to a wide variety of patients with rare medical problems. (J Am Board Fam Pract 2004;17:283–6.)**

Family physicians assume responsibility for the management of undifferentiated problems in unselected patients. They are specialists in the care of common problems.[1] Yet common patients sometimes have rare diseases, and primary care includes the responsibility for recognizing such problems and managing such patients. Studies on rare conditions in primary care are rare. McIntyre[2] highlighted the occurrence of extraordinary cases in his ordinary practice. A few authors discuss the important role family physicians play in the care of patients with specific rare chronic diseases.[3] To help describe the role of rare problems in family practice and the role of FPs in the care of patients with rare conditions, we reviewed the experience in our practice. The goal was not to estimate epidemiologic rates or to measure workload but to identify exceptional cases and investigate the roles played by the FP in the patient's care.

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Methods

Each of 4 FPs in one private practice listed patients with rare conditions. No objective definition of "rare condition" is available to apply prospectively to selection of cases[4,5]; the physicians identified extraordinary cases in the context of their training and clinical experience in community-based practice. Doctors relied on memory aided by appointment records, office staff members, and notes of monthly case presentations. We included only new diagnoses seen in the office. Any diagnosis was included only once; we omitted other patients with the same condition. The author reviewed office records to determine the diagnosis, patient age, and sex, and the roles of the family doctor and consultants in the management of the patient's rare condition. Progress notes, consultant reports, and other medical records established the role of the FP in each step of the process of care. Each patient was scored yes or no for the following questions regarding management of the rare condition. Did the FP identify the problem, establish the diagnosis, provide acute care, provide continuing care, and provide a family context for the patient's care? Did a specialist physician provide consultation

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Table 1. Examples of Rare Conditions Seen in Family Practice by Specialty

Specialty	Condition
Allergy & Immunology	Recurrent anaphylaxis to hops in a brewmaster
Anesthesiology	Recurrent vasovagal sudden cardiac death
Colon & Rectal Surgery	Invasive rectal cancer presenting as elevated prostate-specific antigen
Dermatology	Pemphigus
Gynecology	Ovarian teratoma incarcerated in inguinal hernia
Internal Medicine	
Cardiology	Syphilitic aortitis
Endocrinology	Insulinoma
Gastroenterology	Bleeding ulcer in Meckel diverticulum
Hematology	Erythrocyte aplasia
Infectious Disease	Disseminated varicella
Medical Genetics	Adrenoleukodystrophy with prenatal diagnosis
Nephrology	Polyarteritis nodosa nephropathy
Oncology	Neuroblastoma
Pulmonary Medicine	Catamenial pneumothorax
Rheumatology	Behcet syndrome
Neurological Surgery	Arnold-Chiari Malformation
Neurology	Guillain-Barre Syndrome
Obstetrics	Stillbirth due to true cord knot
Ophthalmology	Scleral metastatic adenocarcinoma
Orthopedic Surgery	Giant cell tumor
Otolaryngology	Horner syndrome caused by neuroblastoma
Pathology	Melanoma presenting as pelvic mass
Pediatrics	Infant botulism
Rehabilitation Medicine	Reflex neurovascular dystrophy
Plastic Surgery	Dermal agenesis
Preventive Medicine	Abnormal Pap smear in male transsexual
Public Health	Pertussis case identifying epidemic
Psychiatry	Obsession-induced carotinemia
Radiology	Inoperable cerebral AVM embolized
Surgery	Carcinoma of the appendix
Thoracic Surgery	Hypoplastic left heart syndrome
Urology	Renal tuberculosis

Examples are drawn from study group of 100 patients.

and/or definitive care for the rare condition? We truncated the list at 100 patients with the most remarkable conditions. Thus, simple frequencies equal percentage of patients with each characteristic.

The study practice comprised 4 residency-trained, board-certified, family physicians with 50 years of experience in aggregate. Over this period, each doctor saw approximately 20 patients a day and attended 20 births a year. The practice is located in a residential, urban community, and provides the full-spectrum of care, including maternity, hospital, emergency department, intensive care unit, coronary care unit, and long-term care.

Results

These 100 patients with rare conditions ranged in age from newborn to 88 years old, with 15 in their first year of life. Mean age for those over 1 year was 38.5 years. There were 54% female and 46% male.

The 100 conditions varied widely across the full range of disease category, affected organ system, and medical and surgical specialty area. Table 1 lists examples, categorized by specialty area. The condition was classified as acute in 33%, chronic in 50%, and developmental in 15% of patients. The condition was life-threatening in 58% of patients.

The family physicians played important roles in the care of the rare condition for most of these

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Table 2. Patterns of Presentation of Rare Problems

Problem Presentation	Clinical Examples
Low incidence or prevalence in practice population	Insulinoma
Classic presentation of a rare disorder	Renal tuberculosis in asymptomatic patient with persistent sterile pyuria
Rare clinical presentation	Melanoma presenting as pelvic mass
Rare clinical course	Cholecystitis in a 5-month-old child
Obscure diagnosis	Diagnosis of carotinemia after extensive studies by multiple consultants for jaundice
Rare coincidence	Sexual assault leading to multiple sexually transmitted diseases and hydatidiform mole
Rare intervention	Fontan procedure for hypoplastic left ventricle syndrome
Rare complication	Ovarian teratoma incarcerated in inguinal hernia
Special family context	Prenatal diagnosis in pregnant woman with adrenoleukodystrophy
Special community context	Diagnosis of pertussis identifying epidemic

patients. The FP was the first to identify the problem in 89%, and established the definitive diagnosis in 54% of patients. The FP provided acute care for the problem in 56%, and continuing care in 76% of patients. Fourteen percent of these patients had been delivered by their family physician. The FP cared for the patient through their final illnesses in 17%. The FP provided a family focus to care that was important to management of the illness for 48% of patients. FPs sought consultation from subspecialty physicians for 85%.

In 62% of all patients, the subspecialists managed treatment of the rare condition. FPs sought second consultations for only 25% of patients. The subspecialties most frequently consulted were general surgery (13), cardiology (10), neurosurgery (9), and neurology (9). Consultation and specialist management were more common in patients with life-threatening conditions.

Discussion

All experienced clinicians have anecdotes about fascinomas, but few reports focus on experience with rare disorders in primary care.[2,3] This simple descriptive study documents that patients with rare conditions can be an important part of family practice and that family physicians can play important roles their care. We drew data from contemporaneous clinical records that are likely to be accurate. The methods, however, have substantial limitations. The study details the experience of one practice in one community. Selecting cases by physician recall may over-represent the most memorable patients, with whom the doctor played an active role. No comparisons are made with other groups of patients or clinicians. It would be interesting to examine the experiences of general internists, pediatricians, and nonphysician primary care clinicians.

This study has the inherent limitations of a case series report. No agreed-upon criteria exist for classification of "rare disease."[4,5] We identified more than rare diseases, however; we studied patients with rare problems as they present to community-based clinicians. Remarkable cases include more than rare diseases. A rare condition may involve one or more of the features listed in Table 2, each illustrated with an example from our practice. Some cases are more remarkable for the context in which they occur. One family physician diagnosed amyotrophic lateral sclerosis in 3 patients in 1 week of office practice. The practice cared for three children in 3 years with craniosynostosis that required surgery. Finding diphtheria in 1 infant led to diagnosis of the disease in 4 generations of the same family.

Only patients with newly diagnosed conditions were included. In some cases, the condition existed before the patient presented, but the diagnosis was made after the patient came into our care. This allows us to describe the role of the FP in identifying and diagnosing these conditions. However, we cannot calculate incidence or prevalence rates. Denominators are always difficult to establish in community-based primary care practices without patient registries. Our recall sample provided no reliable numerators. Without these figures, we cannot determine the rate at which these patients presented in our practices. We did not collect data on visits, tests, or treatments and thus cannot estimate

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the workload or resources devoted to the care of these patients. We did not measure quality or outcomes of care. Further research should address these aspects of the care of patients with rare conditions in other primary care practices and patient populations.

This report discusses only patients seen in the family practice office. We excluded exceptional cases that these doctors have seen in training or in other settings, from botulism and bear bites in Alaska to beriberi and Bilharzia in Zimbabwe.

Family physicians can become experts in the diseases that occur in the patients they serve. If the physician knows the patient that has the disease, the physician can learn about the disease that the patient has. A major source of this education is often the patient, who can become a repository of information gathered over years from many sources: relatives, specialists, publications, and support groups. A patient living in a family with a hereditary disorder is witness to the natural history of its painful progression. A motivated patient armed with such information and insight can be a valuable ally.

This study describes only part of the care provided to these patients. Of course, most had concurrent health care needs. Patients with rare conditions have common needs, too. We have omitted from this report the demographic characteristics of individual patients to protect their identities. We have also omitted the much richer information that is usually known to personal and family physicians. Each of these cases is a human story. This is not just a study of rare diseases or memorable cases but of exceptional patients with special needs that offered us opportunities for professional care and personal growth.

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References

1. Phillips WR, Haynes DG. The domain of family practice: scope, role, and function. Fam Med 2001; 33:273–7.
2. McIntyre FL. One in a million: when extraordinary cases occur in an ordinary practice. J Fam Pract 1993;36:17–8.
3. Atherton A. Primary care for patients with rare chronic diseases. Eur J Gen Pract 1997;3:58–61.
4. Thoene JG, Coker NP, Balistreri WF, et al, editors. Physicians' guide to rare diseases, 2nd ed. Montvale (NJ): Dowden Publishing; 1995.
5. National Organization for Rare Disorders. NORD guide to rare disorders. Philadelphia: Lippincott Williams & Wilkins; 2003.

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