

Zebras on the commons? Rare diseases in general practice

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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 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 38% of patients.

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 38% 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. McIntyre² 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.

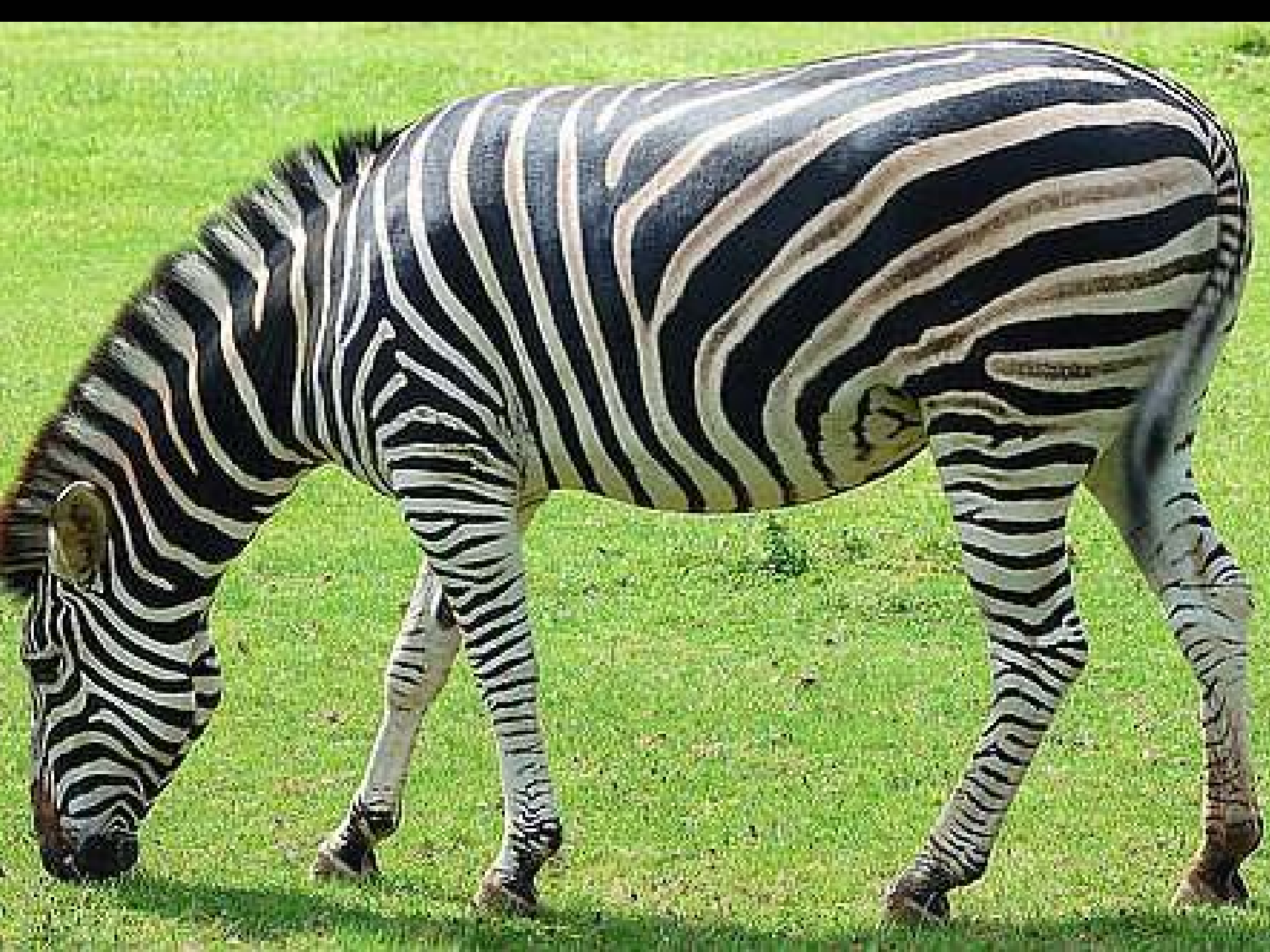
Methods

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 physician 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 memos, 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

When you hear hoofbeats behind you, don't expect to see a zebra.

Theodore E. Woodward, MD
University of Maryland circa 1950

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Jack

Mum

Step Dad

1998

Mood disorder

Little sister

Asthma

Weight

Younger brother

Family conflict

War...

Jack

9.12.87

Difficulty walking by age 3

Duchenne's Muscular Dystrophy

Asthma

13, wheelchair bound, smart

Team – mum, local child and adolescent development unit,
local paediatrician, neuromuscular clinic at CHW, charity
support for family car, computer for home schooling,
powered wheel chair, respite care in the home, respite
care for holidays

Duchenne's Muscular Dystrophy

X linked recessive disorder

30/100000 males

Apparent by age 3-5

Progressive muscular weakness

Contractures

Wheel chair dependent by age 12

Respiratory infections, congestive heart failure

GP's role?

Single diagnoses

6-10% (? 40+ people with rare diseases)

Conditions other than their rare disease

Context – families, communities, schools

GP Role

Diagnose.

Ask more frequently “Could it be a rare disease?” Recognise deviations from common patterns of disease. Be judicious in testing for low-prevalence disorders. Wisely use specialist services for precise diagnoses.

GP Role

Attend to the whole patient.

Provide high-quality care for other health issues including unrelated common conditions and preventive activities (e.g., immunisation, screening and health promotion).

GP Role

Know the disease.

Become knowledgeable about the rare diseases encountered, including natural history, evidence-based treatment options, systematic long-term care, associated problems, and genetics. Seek out appropriate specialist services, international centres of excellence, and local organisations which offer relevant services.

GP Role

Empower the patient.

Encourage patients and their carers to ask questions, and assist them with self-care and decision making.

GP Role

Support the family.

Contribute to the physical, emotional, psychological, spiritual, and social needs of the patient's support network.

GP Role

Advocate.

Support the patient's journey through social service and medical bureaucracies, and interpret written and verbal information.

GP Role

Diagnose.

Attend to the whole patient.

Know the disease.

Empower the patient.

Support the family.

Advocate.

GP Role

D iagnose.

A ttend to the whole patient.

K now the disease.

E mpower the patient.

S upport the family.

A dvocate.

Hopes

Diagnose. Know the disease.

Information and connection...the internet

Diseases, support groups, centres of excellence

Website, 1800 number

Role of primary care

Research with patients and with clinicians

Generic model of rare disease care