

Lesser-spotted zebras

Their care and feeding

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When you hear hoofbeats, think of horses, not zebras." This maxim coined by Dr Theodore Woodward in the 1940s is a useful reminder that we should diagnose the common causes rather than search, at exorbitant length (and cost), for low probability causes. A corollary of this is that rare complications of common diseases are more common than rare diseases. Young doctors at teaching hospitals where rewards come from diagnosing rare diseases must unlearn the habit of searching for them when they stray from the savannas of the wards into the jungle of primary care. Yet "zebras" do exist, and when one arrives, we must know enough that once in a lifetime we can identify the purpuric rash of meningococcal meningitis—or these days, the Koplik spots of measles.

Every primary care practice has a few patients who have been diagnosed with rare diseases—diseases that might occur in 1 per 10000 people, 1 per million, or even less often. But there are many such diseases and each of us will have a different selection. At various times in my own practice I have cared for people with Friedreich ataxia, albinism, type I neurofibromatosis (von Recklinghausen disease), uterus didelphys, specific immune deficiencies, angiodysplasia of the duodenum, and very rare cancers. Some rare diseases are mentioned in the small print at the end of chapters in standard medical textbooks. Others appear only in small print within highly specialized textbooks or are featured only in a few case reports in the literature. These diseases are the *lesser-spotted zebras*: until one arrives in the office, most doctors consider their existence to be as likely as unicorns or hippogriffs.

Caring for zebras

We occasionally identify patients with rare diseases ourselves, often referring them to other specialists for a formal diagnosis, but most zebras arrive with their diagnosis already made, in search of care. Many doctors, especially learners, are inclined to simply refer these patients back to those who made the initial diagnosis, hoping that the other specialists will look after all the needs of these

patients; some feel that the care of zebras is too complex for the front line, or perhaps they lack confidence. They feel overwhelmed by the possible implications and have enough to do caring for the regular basic common problems in their practice: the care of horses. But referral is sometimes impractical. For example, a colleague recently acquired a patient with a disease that has been described in only 5 lineages in the world, and the only physician with experience in treating this disease was located on the other coast of Canada.

Zebras also have all the other medical, psychological, and social problems that other patients present with. They get ordinary common infections and injuries, and develop common chronic diseases, such as obesity, hypertension, diabetes, and heart disease. They need screening, diagnosis, treatment, and care like anyone else. We need to learn the interactions between the unusual medication they use and the other drugs that we frequently prescribe. Expecting a non-family physician specialist will undertake their complete care is neither prudent nor possible, as that doctor might not have the knowledge to treat primary care problems or the systems to do so. Family physicians must distinguish issues related to the disease that actually need specialized care from those we can handle ourselves. Furthermore, if zebras have a disorder that affects more than one system, they might need to see several other specialists, and someone has to coordinate the oftentimes conflicting recommendations. When a zebra is in hospital, whether on a generalist or subspecialist inpatient service, the hospital doctors must also grapple with trying to reconcile conflicting recommendations from various consultants.

Therefore, we should all take the trouble to learn about our zebras. We can read the appropriate chapters in the specialized textbooks, articles from subspecialty-specific journals, or case reports we find in the literature. We can attach copies of any applicable information to the patient's record, either by including photocopies in the file or inserting PDFs or web links into electronic medical records. When a consultation is arranged, we must alert the other specialist about our interest and commitment, so that we can work together. Sometimes we might find ample information on diagnosis but inadequate information on long-term care or a specific issue a patient presents with; so we have to work from basic principles and negotiate with the other specialists about the uncertainties.

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Consultants, on the other hand, assist by identifying the exact species and features of these zebras, and must define for primary care physicians which complications or problems need specialized attention or a different approach from the usual, and what can and should be handled at the front line. Given the range of practice styles, knowledge, experience, and interests among doctors, for each particular patient we might need to negotiate the roles and boundaries between their primary, secondary, or even tertiary health care providers.

Sometimes we need ingenuity to sort out zebra care. The woman with 2 cervixes needs some negotiation with other service providers so, for example, the laboratory does not throw out what appear to be duplicate or mislabeled cervical specimens. After cholecystectomy, the man with neurofibromatosis re-attended the emergency department for recurrent pain. The alarming report of the computed tomographic scan of his thorax and abdomen described the minor biliary leak, as well as many tumours, especially in the pelvis around the prostate, and recommended referral for stage 4 cancer; but the report did not mention the patient's neurofibromas. A request for a re-read, informing the radiologist about this disease, led to a very different amended report, describing the tumours as matching the density of neurofibromas rather than disseminated prostate cancer, to the relief of all concerned. While the neurology textbooks described the diagnostic features and dismal prognosis of Friedreich ataxia, neither they nor the consulting neurologist who saw the patient annually, described how to help this young man live through several years of slow degeneration: long-term palliative care.

The hematologist caring for a man with common variable immune deficiency prescribed frequent courses of antibiotics for sinusitis, which potentially leads to antibiotic-resistant bacteria. When we treated his allergic

rhinitis vigorously and systematically, reducing the swollen mucosa and the exudate in which the bacteria grew, the antibiotics were seldom used.

Rewards of zebra care

Most of my zebras have been wonderful patients; they are very grateful for an inquisitive approach and receiving knowledgeable attention rather than being dismissed as having problems beyond the interest of a mere general practitioner. Once we understand the problems and needs of these patients, they are relieved that they can relate to someone who engages to work consistently with them, even though there is no cure. Then they can stop searching vainly for help from physicians who focus only on their specific disease or only one part of it.

Treating zebras engages the brain, as you work out how to distinguish the unusual features of the disease from the other medical problems that life brings these people. When meeting a zebra, having a sense of curiosity and interest with willingness to search and learn leads to new insights and personal rewards. After the initial effort of learning the issues for this particular one, it is easy to follow along. If they die or move elsewhere, we might never see their particular species again in our practising life, but as other representatives of the genus *Equus* appear we will require new learning. Most hoofbeats do indeed arise from horses, but when zebras turn up, they require equine care that is adapted to their specifics. 🌿

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