

Summary of presentation for:

'Rare Disease Workshop'

## **Zebras on the commons? Rare Diseases in General Practice**

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“When you hear hoof beats behind you don't expect to see a zebra.” This medical aphorism is credited to Dr Theodore Woodward of the University of Maryland in Baltimore. It encapsulates the diagnostic principle that it makes sense to think of the common disease first when making a diagnosis.

However it is now clear that though each individual zebra (rare disease) is uncommon, together they make a mighty herd affecting up to 10% of the community. For the general practitioner there is a paradox. Rare diseases are common.

What is the role of the general practitioner in the care in the lives of people with rare disease? We have developed the following model.

For a patient with a rare disease the GP will:

**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.

**Attend to the whole patient.** Provide high-quality care for other health issues including unrelated common conditions and preventive activities (eg, immunisation, screening and health promotion).

**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.

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

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

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

Australia has the opportunity to test and develop a model for primary care in rare diseases. In particular we need connections so that GP's can rapidly find centres of excellence in Australia and the world. The development of a Rare Disease Plan for Australia is a positive step that will aid general practitioners in their role.