Benefits of Multidisciplinary Clinics for Rare Diseases

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The provision of multidisciplinary services for children (and adults) with rare disorders (encompassing heritable disorders) is an essential feature (*Pillar*) of the management of rare disorders. A number of reports and European technical papers discuss in considerable detail why this is so and the need for a few multidisciplinary clinics in each region which can be "commissioned" as Centres of Excellence. I have chosen to illustrate my presentation by discussing the Connective Tissue Dysplasia (CTD) Multidisciplinary Management Service at the Children's Hospital Westmead. The clinic was started by the senior physiotherapist in 1980 to coordinate diagnosis and management for children with skeletal dysplasias (Disproportionate Short Stature) including Osteogenesis Imperfecta (Brittle Bones). It has become the Centre of Expertise in NSW with contributions from clinical genetics (including genetic counsellors), paediatric rehabilitation, bone & mineral medicine, orthopaedic surgery, physiotherapy, occupational therapy, bone densitometry, orthotics and adolescent medicine. The CTD service provides comprehensive but specialized services for Osteogenesis Imperfecta, Ehlers-Danlos syndromes, Skeletal Dysplasias, Marfan syndrome, Arthrogryposis and Mucopolysaccharidoses. Overall the service cares for children with any one of approximately 600 rare disorders. Grouping the disorders into one of the six major groups is a cost-effective strategy to deliver "therapy" to over 800 current patients. The perceived benefits of multidisciplinary services are: improved patient and parent satisfaction, effective paediatrician or family doctor partnership, improved quality of life and participation in mainstream school. Ongoing evaluation is essential for these programs.