Challenges of transition to adult health services for patients with rare diseases

What can be done for young people stuck in "health care limbo" when they leave paediatric services?

The teenage years are a time of transition, when young people must adapt to enormous physiological and emotional changes but also need time to aspire to the future. Young people living with chronic complex disease have dreams, but their challenges are amplified as they face transition from paediatric to adult health services and begin to take charge of their own complex health care needs.[1]

Young people need the assistance of adult health services to deal with adult issues: sexual health, fertility, drug and alcohol use, mental health, lifestyle-related disease and issues related to disability, employment, education and training. For most of their lives, young people with chronic diseases have been engaged in a paediatric, family-centred multidisciplinary model of care. They need preparation and support to move into adult services, which are more specialised, less integrated, and centred more on the individual than on the family.[1,2] Failed transition leads to poor engagement with health services and adverse health outcomes.[2]

Despite a number of policy initiatives to provide age-appropriate and stage-appropriate care for adolescents and the development of disease-specific transition pathways (eg, for cystic fibrosis, spina bifida and diabetes),[1,3-5] transition is fraught for young people living with chronic and complex diseases, especially rare diseases.

Providing disease-specific clinics for every rare disease is unrealistic; there are almost 10 000 rare genetic diseases alone. Most rare diseases have their onset in childhood, are chronic, complex, disabling and require frequent, specialist care throughout the life span.[6] This necessitates access to multiple doctors, allied health workers, pathology and pharmacy services.[7] Better recognition of rare diseases and increasing survival rates have led to a greater demand for transition services from this group and we must respond to their needs.

Yvonne A Zurynski
BAppSc, MAppSc, PhD,
Associate Professor,[1] and
Deputy Director[2]

Elizabeth J Elliott
MD, MPhil, FRACP,
Professor,[1] and Director[2]

1 Discipline of Paediatrics and Child Health, Sydney Medical School, University of Sydney, Sydney, NSW.

2 Australian Paediatric Surveillance Unit, Sydney, NSW.

yvonne.zurynski@health.nsw.gov.au

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Regardless of which chronic and complex disease they have, these young people face similar problems with the transition to adult care:

- inadequate preparation
- difficulty finding appropriate adult health services
- inadequately coordinated specialist adult services
- unwillingness of general practitioners to take on complex cases
- inadequate resources to coordinate the transition process
- lack of psychological support.

These issues were affirmed in the recent Forum for Young People Living with Rare Disease, attended by 15 young people and 15 parents or carers representing a wide variety of rare chronic conditions: Ehlers–Danlos syndrome, Klippel–Trenaunay syndrome, narcolepsy, cataplexy, Phelan–McDermid syndrome, Duchenne muscular dystrophy, Rasmussen's encephalitis, congenital panhypopituitarism, hypochondroplasia and other skeletal dysplasias.[8]

Forum participants called for:

- comprehensive preparation for transition, involving the family and adult services
- timing of transition according to developmental stage and maturity, not age
- flexibility from adult specialists to allow parents and carers to attend some consultations
- clinics that treat many different rare chronic conditions
- GP clinics that are competent and confident to coordinate care and refer appropriately
- accessible transition coaches or coordinators.

One 18-year-old with a rare syndrome said:

> I'm still transitioning, but it's been a trial. I'm too old for paediatrics but too difficult a case for adult services to treat. I am worried about my health … I don't know who will treat me properly if I end up in hospital.

As most rare chronic diseases are initially diagnosed and treated in childhood, much of the expertise resides with paediatricians, and often there is simply nowhere to transition to. We need to address this imbalance by supporting education on chronic complex diseases in young people — both for medical students and through continued medical education. The ongoing development of the specialty of Adolescent Medicine will support this.

Multidisciplinary clinic models catering for young adults could be adapted to cater simultaneously for many different rare diseases.[5] Such innovative models provide economic efficiencies,[9] facilitate communication among the many health professionals involved in care and ease access for patients. Establishing clinics that involve both adult and paediatric specialists enables sharing of expertise and provides a practical training platform. Incentives beyond the current Medicare rebates are needed to support specialist GP clinics willing to look after young people with rare chronic diseases. Trapeze, a primary health transition service, has been established in New South Wales, although its current focus is limited to diabetes and respiratory disease.[10]

Young people living with rare chronic disease have the right to equitable access to appropriate health care. We need a network of appropriately trained and well resourced transition coordinators to facilitate linkages between young people and health and psychological services and peer support.[8] Evaluation of existing transition services and clinics to inform future service needs should be a priority. Successful transition requires more than a referral letter. It is a *process* that takes time and requires a coordinated system-based approach.

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