



**Forum on transition to adult
health services:
Experiences of young people living with
rare diseases**

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This report is dedicated to the Transition Forum participants who took the time to tell us their experiences and their views, and with careful thought and consideration suggested potential practical solutions to improve the transition journey from paediatric to adult health services.

Acknowledgements

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Executive Summary

Teenagers face many changes and challenges as they transition into adulthood and aspire to the future. Teenagers living with rare chronic and complex health problems face additional challenges as they also transition from paediatric to adult health services. Transition is much more than just a simple transfer of care from the paediatric team to the adult team. It is a process during which young people often require support.

We know little about the transition journey as experienced by young people and families living with rare chronic diseases or medical conditions¹.

The Transition Forum, held on the 23rd of February 2013 at Sydney University, gave voice to young people and their families and enabled them to discuss their experiences, the issues they face, and their needs. Most importantly young people and families were able to identify health service gaps and to provide insightful ideas about how to address these gaps in order to smooth the transition journey. The forum also provided an opportunity to discuss many other issues that youths and families are dealing with during this transition period including access to education and employment and changing relationships with parents, siblings and partners.

The forum comprised a vibrant group of 15 young people and 15 parents or carers who participated in four focus groups; two groups for youth and two for parents or carers. While recognising that health services cannot be provided for each single rare disease as there are thousands of such diseases, the participants suggested a number of improvements:

- Better preparation for transition to adult health services involving the family and allowing adequate time for preparation
- Timing of transition according to readiness to take on a more independent role in their own health care rather than chronological age alone
- Joint consultation involving the paediatric team and the adult team and willingness of adult specialists to involve the family in consultations after transition
- Better integration and coordination in the way adult health services are delivered
- General practice clinics that have a special role in co-ordinating the care of people with chronic complex rare diseases
- A register of general practitioners willing to take on complex and chronic cases
- Better recognition among specialists that the problems people with rare diseases face common challenges regardless of their specific diagnosis
- More resources to coordinate the transition process including moving between health services, access to education and employment, access to benefits, peer support and psychological support.

¹ Zurynski Y & Elliot EJ. Challenges of transition to adult health services for patients with rare diseases. Medical Journal of Australia 2013; 198 (11): 575-576.

Rare Diseases – why are they important?

There are at least 8,000 different rare diseases which affect an estimated 6-10% of the population. That's about 400,000 Australian children. Although each rare disease is by definition infrequent, rare diseases share common features:

- Onset in childhood
- Difficult to diagnose leading to delays to starting relevant treatments
- Most have no cure; are chronic leading to lifelong health problems and disability
- Complex, chronic, requiring multidisciplinary services
- As a group they have significant impact on health and community resources
- Have significant psychosocial consequences for patients and families including isolation and stigmatisation
- Research about the needs faced by the rare disease community is lacking^{2,3}

Why a forum on transition?

Improvements in clinical care for children diagnosed with rare diseases or conditions have led to improved survival rates into adulthood. Consequently the need for transition services from paediatrics to adult services is growing. Despite this, we know very little about the transition journey travelled by young people living with rare diseases. Every February the world celebrates Rare Disease Day (<http://www.rarediseaseday.org/>); a day of action and awareness rising on behalf of all people living with rare diseases. Our Forum contributed to Rare Disease Day activities in Australia.

The aim of the transition forum was to hear from young people and their parents or carers about the issues they faced while transitioning from paediatric to adult health services, any perceived gaps, needs and potential solutions.



² Zurynski Y, Frith K, Leonard H & Elliott EJ. Rare childhood diseases: how should we respond? Archives of Disease in Childhood. 2008; 93:1071-1074

³ Jaffe A, Zurynski Y, Beville L, Elliott EJ. Call for a national plan for rare diseases. Journal of Paediatrics and Child Health. 2010; 46: 2-4

The forum – 23rd February 2013

The forum was organised by the Australian Paediatric Surveillance Unit in collaboration with The Agency for Clinical Innovation Transition Network, Rare Voices Australia and The Smile Foundation, and the TRAPEZE Transition Program.

Opportunities for formal and informal interactions among participants were encouraged through the sharing of stories and opinions in small groups in a supportive non-judgemental environment.

Participants were prompted to discuss the following:

- Their transition journey through health services, identifying good and bad aspects, barriers and gaps and potential solutions
- The challenges of study or work, career development, and access to financial support
- Need for psychological support and peer support during the transition journey
- Changing relationships and developing new relationships.

Thirty people attended the event; 15 young people (age range 15 - 23 years), 13 parents one sibling and one partner. Most were from NSW, including rural centres, two were from Victoria, one from Western Australia. Among the participants were parents of young people who had rare, chronic and disabling diseases, which prevented the young person from attending. A wide variety of rare diseases were represented: Ehlers-Danlos syndrome, Klippel-Trenaunay syndrome, narcolepsy, cataplexy, Phelan McDermid syndrome, Duchenne Muscular Dystrophy, alopecia, Rasmussen's encephalitis, congenital panhypopituitarism, hypochondroplasia and other skeletal dysplasias. As one participant was profoundly deaf, an AusLan interpreter was provided.



Setting the Scene

The first session included presentations about the common challenges that people with rare diseases face, transition services that are currently available in NSW including the Transition Care Network, Agency for Clinical Innovation which facilitates hospital to hospital transition, and the newly established Trapeze Transition Programme which facilitates transition through primary health and community health services (Appendix 1: Transition Forum Program).

Our invited speaker, Catherine Gasparini provided a unique and inspiring talk about her own transition journey as a young person living with cystic fibrosis. Catherine commented on good and bad aspects of her own experience of transition, as she experienced it back in 2005.

The BAD

- Inappropriate accommodation when admitted to adult hospital - sharing with older patients (including men) who have lung diseases and pose a risk of infection
- Older patients in the same ward using bed pans and overnight nebulisers – confronting, noisy and hard to sleep
- Pathology collection – have to wait in pathology (>1 hour) rather than having samples taken in clinic; larger needles (ouch!), no band aids
- Increased costs– have to pay for own parking, food, prescriptions, time off work (standard sick leave allocation is often exhausted as multiple clinic visits and admissions are needed).
- Can't see all specialists in the one clinic, unlike in the paediatric service
- Finding the right GP, someone who knew about CF and was willing to liaise with respiratory specialists took a long time
- Difficult to make decisions about how much to disclose to potential education providers, employers, friends, partners
- Helping education providers and employers to understand the condition
- Difficulties in social interactions; no entertainment; facilities are not age appropriate

The GOOD

- More independence in managing own health – but also more responsibility
- Seeing the doctors solo; finding a really good GP
- Organising own prescriptions
- Being more organised – record keeping of all tests, appointments, admissions, procedures
- Seeking psychosocial support beyond the clinic and family unit – making new friends – including on-line
- Starting romantic relationships



Focus Group Sessions

Participants were divided into 4 focus groups: 2 youth and 2 parent/carer groups. The partner of one of the young women attending and the one sibling attending joined the parent/carer group. Each group had 2 facilitators/scribes. Participants and facilitators were briefed on the topics for discussion and general rules of engagement. These included keeping sensitive information confidential, one person speaking at a time, showing respect for each other's opinions, non-judgemental approach, avoid descending into a "complaint session" but rather, look for solutions. Groups were directed to address the issue of navigating the adult health care system as a priority with other suggested topics or completely new topics if the group felt these were important.

All four groups reported back to the whole forum.

Youth Group Topics

- **Navigating the adult health system – getting the care you need**
- Need for transition coaches or mentors – do they exist? Are they needed?
- Challenges of study or work
- Changing relationships:
 - Your relationship with parents/carers
 - New relationships with friends/partners
 - Relationships with siblings
- Other agreed topics

Parent/Carer Group Topics

- **Navigating the adult health system - What gaps exist in current health services?**
- What services do parents need? Improvements to existing services?
- 'Letting go', dealing with increased independence of your child
- Changing relationships:
 - Your relationship with your child
 - Your child's new relationships with friends / partners
 - Relationships with siblings
- Other topics



Youth Focus Group



Parent Focus Group

Focus Group Findings

While the participants had a variety of rare diseases and medical needs and different levels of disability, many participants were facing similar issues.

Youth Perspective

- Inadequate preparation for transition at the paediatric site
- Need for better “joining-up” of paediatric and adult services
- Adult specialists need to be more open to including parents in consultations initially
- Difficulties finding appropriate adult health services
- Inadequate integration and coordination among adult specialists
- Unwillingness of general practitioners (GPs) to take-on chronic and or complex cases
- Inadequate resources for the coordination/mentoring during the transition process
- Lack of psychological support



Parent Perspective

- Navigating the adult health system difficult, need for more integrated models
- Finding the right GP difficult – need for specialist GP clinics to oversee many complex rare diseases
- Parents need to adjust to their new role in decision-making after transition: need to make hard choices about the balance between allowing child to self-advocate or staying involved
- Administrative issues – transfer of medical records, medicare, access to benefits difficult
- Finding appropriate psychological support difficult, counselling almost never offered



Themes identified by the Focus Groups

Preparation to Transition

Participants described a lack of preparation for transitioning from paediatric health services to adult health services. While there is currently no consensus in the literature as to the most appropriate age to begin transition, all participants agreed that **preparation** was the key for a successful transition and having appropriate information available early on, well before transition actually occurs, was paramount. Most participants felt that meeting the new medical team in the adult health service before the transition actually occurred was very important to enable young patients to become familiar with the new health setting and to meet their new health providers.

“...no one informed me that I needed to transition until a month before I turned 18, so essentially it was thrown at me and I’ve been in limbo ever since.” A young woman with Klippel-Trenaunal Syndrome.

Focus group participants suggested that a ‘transition pack’ containing relevant documents, fact sheets, tips and advice written in language easily understood by a lay person be made available at paediatric health services prior to transition and at adult health services after transition, as well as on hospital websites. Although the NSW Agency for Clinical innovation, Transition Care Program, provides similar packs, these had not been made available to clinicians involved in transitioning the young patients participating in the forum. Wider distribution and awareness raising about existing transition services and resources was identified as a priority.

Accessing General Practitioners

Finding a suitable General practitioner was a common issue raised by both parents and youth focus groups. Participants commented that many GPs were unwilling to take on such complex cases, with many GPs not having the knowledge or confidence to deal with rare diseases/ conditions.

“I see several GPs but each knows something but none knows everything”.

“I tried finding a good GP. I saw 11 of them... and then gave up.”

Many of the forum participants had been to see many different GP's. Participants told us that many required double bookings with their GPs to enable sufficient contact time which added to costs as most GPs now charge an additional fee for service in addition to medicare rebates, for consultations.

The group suggested that GPs should have incentives such as a “GP management plan for rare complex disease” allowing GPs to spend more time with patients living with a rare complex disease or condition. Although GPs have access to “chronic care plans” for their patients these plans are designed mainly for older adults with more common and well known chronic conditions such as diabetes, heart disease, chronic obstructive pulmonary disease etc.

Forum participants acknowledged that it is impossible for GPs to know about all of the rare diseases as there are simply too many. However, they wanted GPs to have adequate time to access education and available resources about rare diseases and to learn about rare diseases when such patients present to them. They also called for specialist rare disease GP networks of clinics staffed by GPs who are willing to coordinate care and to refer appropriately.

The newly established Trapeze Transition Service in NSW aims to smooth the transition journey through primary care by providing case coordination and sourcing appropriate services in the community. Trapeze has enormous potential to address the difficulties that patients with rare diseases face when attempting to use primary care, however, for the moment Trapeze services are limited to supporting young patients with diabetes and respiratory conditions.

Experiences of Adult health services

Need for integration and coordination

Many of the participants commented that there were limited links between paediatric and adult services. The overall perception from the groups was that the adult health system is disjointed and uncoordinated, made up of '*specialised silos*'. Participants called for a more multidisciplinary approach to providing healthcare for chronic and complex diseases.

"The respiratory specialist will only deal with lungs and nothing else."

"The adult specialists don't talk to each other."

"We have to repeat our medical history to every single doctor we see."

Participants also raised the issues of long waiting times for specialist appointments, for some this was six months or more.

"What am I supposed to do when I'm worried about my health now?"

Some participants had tried seeing specialists in the private sector which enabled them to be seen more quickly, though the financial costs were prohibitive. Most participants indicated that they wouldn't be able to afford fees charged by doctors working in the private sector.

"Even though we have a psychiatrist in our [regional city] he has a very long waiting list. He is a private psychiatrist and the medications he prescribed are not covered by PBS. He is expensive as are the medications." Mother of a young woman with Phelan Mcdermid syndrome.

Many participants raised issues of equitable access to appropriate health services stating:

"You have to get lucky to get the right doctor"

"You have to FIGHT for the care you need and deserve"

"It shouldn't be like this – access to appropriate health care is a basic human right"

Nowhere to go: The need for appropriate services in the adult health sector

There is an imperative for paediatric services to transition young patients to adult services by the time they are 18 years old. Some participants indicated that there were simply no adult services available for them to transition to.

“He’s been waiting for transition but they don’t know where to shift him. He is too old for paediatrics. But where can he go when he’s pretty much lived in a paediatric hospital all of his life.”

“I’m a hot potato.”

“I’m too old for paediatrics, but too difficult a case for adult services to treat in this country. I am so thankful to all the people who are helping me with this journey. I cannot fault them at all for how they’ve tried to get me into adult services. I wish it was easier for them.”

Participants also brought up problems with the healthcare workforce including:

Highly mobile health workforce where highly skilled allied health and nursing staff move away from direct patient care.

“Just as you find someone who knows about your rare disease they move on! They go to administration where they seem to get paid more.”

Participants felt that much of the care provided by clinicians who are expert in rare diseases was initiated by the rare disease expert and not well supported by the health system. As rare diseases are rare, and so are the experts in rare diseases. These experts are difficult to replace.

“Continuity with doctors and specialists is often really hard because they move on and often months or years of work is undone.”

For some, being placed in wards with elderly patients had been distressing.

“As I get admitted to the Neurology ward, it has been distressing to be exposed to very elderly stroke or dementia patients who tell you they just want to die.”

One 20 year old young woman had been seeing a paediatric rheumatologist since she was three years old and she simply said:

“I don’t want to transition; I continue to have a paediatric rheumatologist.”

Financial Impact

The financial burdens for patients and their families were a significant issue with costs associated with numerous GP appointments, specialists, allied health professionals, hospital admissions and medication costs.

Some participants told us that they didn't qualify/ didn't fit criteria for assistance; forms for financial assistance were inflexible and failed to recognise people with rare chronic conditions:

“you can't tick the boxes if you don't fit the boxes”. The process to access financial benefits is complex and difficult: *“The paper work is brutal.”*

Participants suggested workshops to assist with navigating the different financial schemes e.g. Medicare, Centrelink, Disability Support Pension.

Psychological Support/Support Groups

One of the themes that was raised by both parents and youth groups was the lack of psychological support during the transition process. Participants noted that it was a rare occurrence to be referred for psychological support with many having to ask for this service.

Some participants sought support from peers where possible but many had to look overseas to find support groups or use online forums/blogs. One of the highlights from this meeting was that participants vowed to establish a support group via social media.

Other proposals from the forum participants consisted of forums and support groups that were age and sex based, available 24/7, promoted self-education and were general for any rare disease rather than for specific diseases.

“We feel isolated, like we've been swallowed up into a void”

“I had to ask to get help from a psychologist”

Changing Relationships

Parents and Children – letting go

Many parents face the difficulty in 'letting go', during transition and they indicate that it can be hard to find the right balance between allowing the child to self-advocate and to become more active in their own health care or to stay involved where necessary. For some parents this can be more difficult if the young person is unable to self-advocate due to significant physical or intellectual disability. Parents felt that they were seen as demanding; one parent stated that she was labelled as an "overbearing mother". This despite the fact that her son has multiple chronic and complex health issues stemming from a very pre-mature birth:

"I feel like I'm some giant octopus trying to hold on to make sure he gets the right health care."

"Parents are usually pushed away when the kids reach 18 because they have legally become adults. Many of the kids still need parental support and involvement. The system does not have all the answers and very few services".

Differences in gender roles of parents were discussed in one group. The fathers felt that they provided support to their children in a different way than mothers. Fathers were more likely to support their child with a rare disease by providing diversions through, play, games and outings whereas mothers took on the more practical carer role by providing care and support needed for activities of daily living.

Young people also noted that it can be difficult seeing a GP/specialist on your own for the first time. *"it can be a shock when you see a doctor on your own"* ; *"you need to be confident to do so"*. One of the solutions offered by the group was for parents to still be involved in GP and specialist visits *"see the young person first and then bring parents carers in"*.

Siblings

The impacts on siblings of children and young people affected by chronic complex diseases are well recognised. Siblings often miss out on parental attention, on outings and resources as the family has to focus their care and attention on the affected child. Siblings often provide practical help in the care of their sibling; however, these relationships change as the affected child becomes a young person in their late teens and wants to assert their independence. It is difficult for siblings to adjust who may interpret this new independence as rejection.

"My sister is older than me – she doesn't want my help"

Employment

Employment was a key theme that was raised by participants. Participants discussed the idea of disclosure to employers regarding their rare disease/condition, issues regarding sick leave and long gaps in employment due to illness, the need for more transferable skills, difficulties in working and studying and still being able to cover medical costs. Many felt that there is no incentive for young people to work as the disability support pension is cut off as soon as the young person is employed. Often young people are worse off when employed as they lose rebates and their expenses for medications and services increase. Participants called for a health care card for all people with rare chronic and complex diseases and financial support to cover the frequent and complex medical care they require at least until they are earning an average wage.

“My plan is to marry RICH.”

“There’s no incentive to work. I can only work part-time because of my condition. When I work I lose my benefits. I can’t afford all the drugs and appointments.”

We need to change “the SYSTEM”

Although the focus groups concentrated mainly on transition through the health system, other relevant problems related to social, educational, employment and financial supports were also discussed. The main themes were: complexity, lack of coordination across sectors and lack of recognition of the needs of young people living with rare diseases.

“Schools do not care for kids with rare conditions. Students families and teachers need a lot more education about tolerance, acceptance and equality.”

Participants expressed frustration at the lack of integration between health, education services and social welfare services. They felt that the system was geared to support well-known chronic diseases and conditions such as heart disease, diabetes, COPD and cancer but young people with rare and less well known diseases fell between the cracks.

“Families have to fight to get basic health, educational, employment and social services.”

The groups touched on the National Disability Insurance Scheme (NDIS). They welcomed NDIS which they hoped would provide services and support to people according to their disability and needs and not according to a diagnostic label.

Participants identified a general lack of recognition by the current systems of the impact of their rare condition on their health, psychosocial functioning and wellbeing and their financial situation.

“Not all doom and gloom. Many liked the independence of adulthood and having their own money, driving, relationships and travel. Not all can do this, even as adults.”

Summary of recommendations by Participants

Young people need adult health services to independently deal with various adult issues including sexual health, fertility, drug and alcohol use, mental health, life-style related disease, and issues around disability, employment and education. For most of their lives, many of these young people have been engaged in the paediatric family-centred setting. Young people and their families need preparation and support to move into adult health services which are more specialised, less integrated, and centred on the individual rather than family. Failed transition can lead to poor engagement with health services and adverse health outcomes. Transition services need to be available to a wide age range, there should be no set age for transition due to issues with delays in diagnoses for many with rare conditions.

Transition issues facing young people with complex, chronic disease are similar no matter which disease they have:

- Inadequate preparation
- Difficulties finding appropriate adult health services
- Inadequate coordination among adult specialists
- Unwillingness of general practitioners (GPs) to take-on complex cases
- Inadequate resources for the coordination of the transition process
- Lack of psychological support

Where to from here – Participant Recommendations

We need to improve transition services from child to adult health because there are thousands of rare diseases, most in childhood, that are chronic, complex, often disabling and require frequent, specialist care throughout the life-span. Better recognition of rare diseases and increasing survival rates have led to a greater demand for adult services for this group.

Providing disease specific clinics for each rare disease is unrealistic. Rather, services should be established for broad disease categories with similar health needs. The needs for access to appropriately coordinated services able to deal with complex cases and disability, access to equipment and allied health services and need for psychosocial support are common to many rare chronic diseases. A multidisciplinary service would simplify their care, increase efficiency of service delivery and save resources.

Forum participants called for:

- Comprehensive preparation involving the family and adult services
- Timing of transition according to developmental stage and maturity, not age
- Flexibility in consultations with adult specialists allowing parents/carers to attend some of the time
- Clinics that can manage people with a range of treat rare chronic conditions rather than special clinics for individual diseases
- GPs or GP clinics that are confident to coordinate care and refer appropriately , and willing to learn about rare chronic conditions
- A “one-stop –shop” where transition coaches/coordinators are easily accessible and could provide information and resources for young people
- On-line support group to provide peer support and share information

***We would like to thank all the participants at the forum
for their valuable input and enthusiasm on the day.***



Participant's Experience – their transition Journey

Prior to the Transition Forum, participants (youth only) were asked to write down their story to help us understand their experiences of transition. Youth were provided with a form containing a number of prompt questions but were asked to talk about any aspect of their transition journey. The prompt questions included:

What challenges have you faced because of your medical condition?

How would you make things better?

If you have transitioned to adult services tell us about your transition journey.

Tell us about your expectations for transition.

You can talk about any aspect of transition: health, education, relationships, information needs; support needs and your feelings about how transition could be improved.

Siobhan's Journey

18 year old Siobhan has Klippel-Trenaunay Syndrome (KTS). Siobhan is currently transitioning from paediatric to adult health services.

What is KTS?

KTS is a rare congenital condition; the cause of KTS is unknown.

There is no cure for KTS and treatment is symptomatic.

KTS is a progressive disorder, complications may be life threatening

The condition affects individuals blood and lymph vessels, there is abnormal growth of soft and bone tissue.

"I haven't been able to complete school properly. I've been attending school on and off for the last eight years with varying degrees of success, most of this is due to surgery, pain and fatigue. I am at TAFE completing a certificate IV course so I can go to University.

I have trouble keeping friendships / relationships. Again, I didn't attend school properly so I have no long lasting relationships from that, and most people don't understand and seem confused by my illness. It also strains my family relationships and I feel bad for my family who have to listen to me when I'm feeling ill. They can't help me, and I feel terrible for it.

I've had an abundance of surgeries, some terribly serious and some only slightly so, some that weren't meant to be serious but somehow ended up costing me years of my life.

I'm still transitioning, but it's been a trial. As I have a rare condition it's almost impossible to find doctors who will take on a case like mine. Added to the fact that no one informed me that I needed to transition until a month before I turned eighteen, so essentially it was thrown at me and I've been in limbo ever since. I'm too old for paediatrics, but too difficult a case for adult services to treat in this country. It's tiresome really. I wish there was a simple way for this to go. I wish I had a simple illness to work with".

Eleanor's Journey

Eleanor is a 21 years old with narcolepsy and has already transitioned to adult health services.

What is Narcolepsy/Cataplexy?

Narcolepsy is a chronic disorder of the Central Nervous System, characterised by the brain's inability to control sleep-wake cycles.

There are several causes; one known cause is lack of brain neurotransmitter called hypocretin.

Narcolepsy affects both males and females equally. It often starts in childhood /adolescence and is lifelong

"Narcolepsy has caused a lot of issues in my life, but a lot of them have gotten much easier since I was diagnosed and started taking medication. When I was still in high school learning was quite difficult as I would fall asleep in at least half my classes every day.

Narcolepsy has also caused mental issues like mood swings, being constantly tired makes you completely on edge and unable to deal with even small problems that might come up in everyday life. I also didn't really go out much or do many things, this made me even more frustrated because I was sick of doing nothing but knew that if I tried to do things I'd probably just fall asleep anyway. Another big

challenge has been that one of my symptoms is cataplexy, which makes me lose muscle tone, mostly when I laugh. To a lot of people it is funny but for me it's really embarrassing as people really do think that you must be drunk or mentally disabled, as you can't laugh without having to brace yourself against something so you don't fall over and you can't keep your eyes open properly.

No-one really knows what narcolepsy is, or they have only seen a ridiculous exaggerated version in a movie once. This makes it really hard to get people to understand how much of an impact it can have on your life. I think raising awareness in the general population would be a good way to make some improvements to the issues of understanding from others, as well as possibly helping to promote earlier detection and diagnosis. I think if I had been more aware of narcolepsy and what the different symptoms are; it would have been obvious to those around me that I might be suffering from it.

When I was diagnosed, I wasn't made aware of the fact that people with Narcolepsy are far more likely than the general population to suffer from depression and other mental illnesses, as well as other issues such as avoiding social situations, feeling isolated and misunderstood etc.

For me, doing more research and finding out that these feelings were actually completely normal for a person with narcolepsy has made it so much easier to manage, as I no longer feel like I "should" be able to just deal with it. I have even tried to share this sort of discovery with others; I completed my Bachelor of Design last year and for my final project I designed a book filled with information that could be easily read and understood by people with narcolepsy, as well as acting as an aide to help their friends and family understand their condition.

I've joined an Australian Narcolepsy Support Group on Facebook which is really helpful; people just ask questions or talk about common experiences. It's comforting to talk to other people who understand your issues in a completely different way than your friends or family ever could. I also like the chance to be able to help out other people with their questions and give advice when I can".

Evaluation

A post forum evaluation was conducted, participants were asked to complete a short on-line survey regarding the event.

A total of 17 surveys were sent out, 11 (65%) of participants responded to the survey. Six of these were young people (age range 15 to 22 years) and 5 were parents.

Overall 91% of respondents enjoyed the forum. The majority of respondents believed that the topics covered in the morning session were very relevant or relevant (90%), with only 1 participant finding the topics in the morning session not relevant at all.

Overall, participants felt that the focus group facilitators were excellent (73%) or good (27%).

When asked what they enjoyed most about the forum, participants stated that they most enjoyed the focus groups and the opportunity to meet other people with rare diseases, followed by the presentations and lucky door prizes. All respondents said that they would attend similar events in the future.



Transition Packs from Agency
for Clinical Innovation (ACI)

Appendices

1. Forum Program

Forum for Young People Living with Rare Disease: The challenges of the many transitions to adulthood!

Saturday 23rd February 2013
University of Sydney, New Law School Building

Programme

Aims:

To document the issues experienced by young people living with rare diseases when transitioning from child health services to adult health services.
To develop advocacy tools proposing changes or improvements in transition services.

Expected Outcome:

A report summarising the forum findings will be published. The report will be made available to service providers, government agencies, rare disease organisations and the rare disease community, and will support advocacy for improved transition services.

Forum organisers: Australian Paediatric Surveillance Unit
Rare Voices Australia
ACI Transition Care, Agency for Clinical Innovation

Time	Title	Speaker/Facilitator
8.30-9.00	Coffee/registration	
9.00-9.05	Welcome	A/Prof Yvonne Zurynski
9.05-9.15	Rare diseases – unique needs	Prof Elizabeth Elliott
9.15-9.45	Transition services in NSW	Ms Lynne Brodie, and Ms Jude Foster
9.45-10.00	The need for family support and advocacy	Ms Tam Johnston CEO SMILE Foundation and Ms Megan Fookes RVA
10.00-10.20	My Transition Journey with Cystic Fibrosis	Ms Catherine Gasparini
10.20-10.40	Morning tea	
10.40-10.50	Briefing for breakout group discussion	A/Prof Yvonne Zurynski
10.50-12.30	Group discussions	*Facilitator for each group
	4-5 break-out groups	
12.30-1.00	Lunch	
1.00-2.00	Feedback from Groups	
2.00-2.30	Summing up	Prof Elizabeth Elliott

Breakout group discussion will include but will not be limited to the following:

- Navigating the adult health system – getting the care you need
- Need for transition coaches or mentors – do they exist? Are they needed?
- Challenges of study or work
- Relationships – with parents, friends, partners

Proudly supported by: Australian Paediatric Surveillance Unit; Rare Voices Australia; Smile Foundation; The University of Sydney; and Transition Care, Agency for Clinical Innovation



2. Convenors and Organisers

Yvonne Zurynski, Deputy Director, Australian Paediatric Surveillance Unit and Associate Professor, Discipline of Paediatrics and Child Health, The University of Sydney

Lynne Brodie, NSW Transition Care Network Manager, Agency for Clinical Innovation.

Imogen Yang, BEECHAC Project Coordinator, Rare Disease Day Sydney Working Group member, (former board member Rare Voices Australia), independent Rare Disease advocate

3. Speakers and Facilitators

Catherine Gasparini (Invited Speaker)

Catherine Gasparini is a 26 year old living with, and trying to conquer, Cystic Fibrosis. She completed her Bachelor of Arts (Psychology)(Honours) degree several years ago and loved uni so much she never left. Catherine now combines her passion for psychology and health matters by working as a Disability Advisor at the University of Western Sydney. In this role she supports current students with disability, chronic illness and mental health conditions through their degree. In addition to paid employment, she considers the daily regime of managing her Cystic Fibrosis another job in itself!

Lynne Brodie (Speaker & Facilitator)

Lynne is a registered nurse with qualifications in paediatrics, disability nursing, health management, project management and a BA in psychology. The majority of her nursing career was spent at the Royal Alexander Hospital for Children in Sydney where she held a variety of positions including nurse educator, burns CNC, Nursing Unit Manager and Manager of Clinical Operations. Lynne moved to her current role as NSW Transition Care Network Manager with the Agency for Clinical Innovation in 2004.

Elizabeth Elliott (Speaker & Facilitator)

Professor Elizabeth Elliott AM is Founder and Director of the Australian Paediatric Surveillance Unit; Consultant Paediatrician at the Children's Hospital Westmead; an NHMRC Practitioner Fellow; and past Convenor of the International Network of Paediatric Surveillance Units. She holds NHMRC and ARC Grants relating to rare diseases and has published widely in this field. She is involved in the diagnoses and management of rare diseases; is on the SMILE Board and Steve Waugh Foundation Medical Committee and is an advocate for people living with rare diseases, including through development of a National Plan for Rare Diseases and involvement in World Rare Diseases Day.

Jude Foster (Speaker)

Jude Foster is the Manager of the new state wide Trapeze Adolescent Service at the Sydney Children's Hospitals Network. In 2006 Jude was awarded a Churchill Fellowship for the development of the Wrap Around Kids program which was established in 22 Australian schools in NSW and Victoria. Wrap Around Kids was awarded a National Quality Use of Medicine Award in 2003 by the National Prescribing Service and was recommended as an intervention by the NSW Department of Education Inquiry into Early Intervention of Learning Difficulties. Most recently Jude was the Chief Executive Officer at miVitals Technology Pty Ltd. Jude was President of the Learning Difficulties

Coalition NSW, the peak advocacy body for families of children with learning and attention difficulties for 10 years.

Imogen Yang (Facilitator)

Imogen Yang is a co-founder of BEECHAC Inc - the Bladder Exstrophy Epispadias Cloacal Exstrophy Hypospadias Australian Community; a member of the Rare Disease Day Working Group and joint coordinator of the Sydney Rare Disease Day campaign in 2012, and a founding board member of Rare Voices Australia and inaugural editor of the Rare Voices newsletter. She is active in developing collaborative projects between the health and philanthropic sectors and other rare disease support organisations. Imogen works across a number of intersecting fields including arts and culture, community cultural development, media & communications, disability advocacy, drug and alcohol support. She believes in the ability of individuals to bring about change in society. Her lived experience with rare disease is through family relationships and as a partner, and through this has formed a commitment to promoting the importance of both psychological and physical health for overall well-being.

Tam Johnston (Speaker)

Tam has spent the last nine years with the National Australia Day Council, most recently as National Program Director and Deputy CEO. In this role Tam was responsible for the Australian of the Year Awards as well as Australia Day celebrations nationally. Tam has also worked in Public Affairs for the Australian Government and as a Public Relations consultant in private industry. She holds a Bachelor of Communication, a Graduate Certificate in Management and a Masters of Business Administration. *"I am delighted to join the SMILE team as CEO and look forward to the opportunity to help support vital research into rare diseases and support families when they need it most."*

Megan Fookes (Speaker & Facilitator)

Megan, who was born in Victoria, married with 2 children and resides in Sydney, a former Primary Teacher both in Victoria and NSW. Her professional association with Rare Diseases stems from a very personal connection. Her late father waited 48 years to receive a diagnosis of a rare condition called Fabry disease. Her parents who were very keen to learn more formed a patient centred support group; Fabry Support Group Australia (FSGA) 19 years ago. She has been actively involved in the group for the last 15 years. FSGA is also a member of another important organisation called FIN (Fabry International Network) a global, independent network of Fabry patient associations whose purpose is to collaborate, communicate and promote best practice to support those affected by Fabry Disease in the global community. FIN represents over 27 Fabry Patient Organisations in over 24 countries. Megan has been actively involved heading the Board of this international organisation. Recently she has joined a newly formed national organisation called Rare Voices Australia (RVA) working as their Acting Executive Director 3 days a week. RVA established following a call from over 200 attendees at the 'Awakening Australia to Rare Diseases' international symposium in Fremantle in 2011. Megan is keen to utilise all her experience and knowledge to help RVA be the voice for ALL Australians living with a rare disease.

Marie Deverell (Facilitator)

Dr Marie Deverell is a senior research fellow who joined the APSU in January 2012 from Western Australia. Marie has a PhD in Paediatrics and Child Health. Marie has worked in various health areas including respiratory research, health promotion, advocacy and health policy. She is currently working on a number of rare disease projects, including a review of transition services.

Yvonne Zurynski (Speaker & Facilitator)

A/Prof Yvonne Zurynski is Deputy Director of the APSU and Associate Professor Discipline of Paediatrics and Child Health, The University of Sydney. Yvonne has a PhD in Medicine and a special interest in rare diseases research and epidemiology. She is interested in the experiences of patients and families living with rare disease and currently leads an Australian Research Council project in this area. She sits on the Medical Health Advisory Committee of the Steve Waugh Foundation and works collaboratively with the SMILE Foundation, AGSA and other parent support organisations.

4. List of Resources

Resources and useful Links:

- ACI Transition Care Program - <http://www.aci.health.nsw.gov.au/networks/transition-care>

Contact ACI - ACI reception on (02) 9464 4666 or Email: info@aci.health.nsw.gov.au

- Trapeze Transition Service - *Contact : Jude Foster* (Manager), Tel: (02) 8303 3600 or Email: Jude.foster@health.nsw.gov.au

- Chronic Illness Peer Support (ChIPS) - <http://www.rch.org.au/chips/>
Contact ChIPS – (03) 9345 6616

- Siblings Australia - <http://siblingsaustralia.org.au/>

- Carers Australia - <http://www.carersaustralia.com.au/>
Contact Carers Australia – (02) 6122 9900

- Rare Voices Australia (RVA) - <http://www.rarevoices.org.au/>
Contact RVA - (02) 9967 5884 or Email: info@rarevoices.com.au

- Smile Foundation – <http://www.smilefoundation.com.au/>
Contact Smile – (02) 94094860 or Email: info@smilefoundation.com.au

- Steve Waugh Foundation (SWF)- <http://www.stevewaughfoundation.com.au/>
Contact SWF – (02) 9964 6255 or Email: info@stevewaughfoundation.com.au

- The Association of Genetic Support of Australasia (AGSA)- www.agsa-geneticsupport.org.au
Contact AGSA – (02) 9211 1462 or Email: info@agsa-geneticsupport.org.au

- National Disability Insurance Scheme (NDIS) - <http://www.ndis.gov.au/>
- Orphanet - <http://www.orpha.net/>
- National Organization for Rare Disorders (NORD) - <http://www.rarediseases.org/>
- Higher Education Disability Education Support Program - <http://www.innovation.gov.au/HigherEducation/Equity/HigherEducationDisabilitySupportProgram/Pages/default.aspx>

5. References

1. Zurynski Y & Elliot EJ. Challenges of transition to adult health services for patients with rare diseases. Medical Journal of Australia 2013; 198 (11): 575-576.
2. Zurynski Y, Frith K, Leonard H & Elliott EJ. Rare childhood diseases: how should we respond? Archives of Disease in Childhood. 2008; 93:1071-1074
3. Jaffe A, Zurynski Y, Beville L, Elliott EJ. Call for a national plan for rare diseases. Journal of Paediatrics and Child Health. 2010; 46: 2–4

6. Organisations who supported the forum

The Australian Paediatric Surveillance Unit (APSU)

NSW Agency for Clinical Innovation Transition Care Network

Rare Voices Australia

The Smile Foundation

The University of Sydney

