

Summary of presentation for: 'Rare diseases Workshop'

## **Helping families affected by rare diseases**

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Families who receive a diagnosis of a significant, chronic disorder are forced to live a life they did not plan for. It is not usual for human beings to choose and sustain long-term the most difficult of all their options.

It is reassuring that despite intense grief and difficulties families have the resilience to find and live their new life, with redefined expectations. Our families really want to live life as if death is not an imminent part of their life.

They do need help from people, services, systems and technology that they, for the most part, did not previously know existed. Help from different sources is needed for every member of that family. Where do we start, who do we enlist, how do we flexibly meet their evolving set of needs?

This presentation will explore the medical, emotional, and practical help our families need, which I will try to illustrate by relating to a 'real family'. A multidisciplinary team approach is very powerful – but still – we are often left feeling 'how did we really help this family – it is just not enough!'

Some of the specific areas for support are : genetic; social; education; travel; accessing tertiary services; mental health; therapy; transition and end of life. Each of these issues is multifaceted and variable for each different family and individual.

We learn from our families and improve the quality of our 'help' – and families learn to accept help and improve the quality of their hope.