

## CONFERENCE ABSTRACT

# Hearing the lived experience of young women with a rare genetic disorder 22q11.2DS regarding integrated care

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**Introduction:** The lived experience of young people with chronic disease has seldom been heard in health research despite the clear policy imperative to hear the voices of children and young people as service users and rights holders. This participatory research sought to redress this by empowering young women with 22q11.2 Deletion Syndrome, a rare genetic disorder which can cause chronic, multiple and complex health difficulties, to express their lived experience of transitions from paediatric to adult care.

**Methods:** Radically different from the old paradigm of positivist science, the transformative research methodology 'Participatory Action Research' (PAR) was chosen to empower participants by conducting research 'with' and not 'on' them, validating their lived experience and ability as service users to inform service provision and policy.

Six young women with 22q11.2DS aged 21 to 35 years were recruited to a 'Young Experts by Experience Panel' (YEPP). Arts based, narrative methods including body map drawings, lifelines and photo-voice were employed to initially elicit qualitative themes. Subsequent focus group discussions explored these themes. Digital story methodology informed a short video highlighting the YEPP's experience and recommendations regarding an integrated care clinic for children and young people with rare genetic disorders.

**Results:** Research participation outcomes included enhanced health literacy, acceptance of diagnosis, peer support, self-advocacy, confidence, autonomy and a sense of belonging to a unique community. Emerging themes included acceptance of chronic illness, self-care management of mental health and wellbeing and the challenge of communicating with multiple care providers complicated by mild intellectual disability and speech and language difficulties. Identified health service needs included the need for an integrated care coordinator and an integrated multidisciplinary care clinic to support with the transition from paediatric to adult healthcare.

**Discussion:** The young adult service users specified their preference for a 'one stop shop' to provide integrated care across their lifespan. They reported that the current absence of integrated care causes anxiety and fear as participants struggle to cope with explaining their rare disorder to healthcare providers, to communicate effectively with clinicians from multiple disciplines and with their reluctant dependence on ageing parental carers to facilitate access to appropriate healthcare.

**Conclusions:** Integrated care provision was identified by participants as the pathway to greater autonomy, self-care, successful transitions and positive health outcomes.

**Lessons learned:** This participatory approach to research was an empowering process for young adult patients which resulted in a compelling request to policy makers to enable integrated care for rare chronic diseases. Additionally producing a visual digital story of the research process facilitated dissemination of young people's voices to be heard in multiple settings, including clinical, academic, community and policy environments.

**Limitations:** Notwithstanding the unique richness of the qualitative findings into the lived experience of young adult female service users, the research was not informed by the male young adult experience of their chronic disorder due to lack of available male participants.

**Suggestions for future research:** Further participatory research with children and young people with chronic rare diseases and their parents could usefully inform the development of an integrated care pathway as a model for chronic rare diseases. Such research should strive to include the perspective of both genders' perspective.

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**Keywords:** participatory action research; young experts by experience; rare genetic disorders; 22q11.2ds

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