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CONFERENCE ABSTRACT

Developing integrated care in the context of rare chromosomal conditions: 22q11 Deletion Syndrome; A parent/clinician collaboration.

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Introduction: 22q11.2 Deletion Syndrome is the most common rare chromosomal disorder after Down's Syndrome with a prevalence rate of between 1 in 2000 and 1 in 4000 live births. 22q11.2DS is characterised by a wide range of complex medical, developmental and mental health problems. Over 180 physical, functional and psychological associations have been described. The cost of care to families and to the health system is high because the intervention of multiple specialist medical and psychiatric services is required over the course of a lifetime as well as ongoing educational and psychosocial support. Costs are increased when this care is provided in a fragmented way without interdisciplinary co-ordination. The key recommendation from the European Union Committee of Experts on Rare Diseases is multidisciplinary care of rare disease patients at Centres of Expertise including psychosocial care and the development of healthcare pathways. However neither a centre of expertise nor an integrated care pathway for rare diseases have been developed in Ireland to date.

Short description of practice change implemented: 22q Ireland Support Group as a patient organisation, with the support of an independent research consultant, initiated dialogue with a large group of specialist clinicians from a range of health disciplines and institutions. The practice change outcome has been the establishment of a core patient-clinician working group who have collaboratively developed a comprehensive business case to seek political support, statutory and research funding for the development of the multidisciplinary networked model of integrated care. A care pathway planning day was also facilitated to develop a recommended coordinated care model pathway specific to the Irish health service context.

Aim and theory of change: The shared aim is to develop a clinical network, center of expertise and care pathway for 22q11.2DS informed by carers and service users, building upon existing specialist expertise within the health system. To achieve this, a clinical care coordinator, a lead paediatrician and a family support coordinator will work with service users, families and clinicians to develop and co-ordinate the care pathway in a multidisciplinary network of expertise.

Targeted population and stakeholders: Children and young people with the rare genetic disorder 22q11.2 Deletion Syndrome; Carers

Timeline: This patient driven collaborative project began in June 2016 and is an ongoing initiative.

Highlights: (innovation, impact and outcomes) This patient driven Innovation involves carers, service users, clinicians and researchers as equal partners in the development process. Impact includes securing clinical commitment and seed research funding from the Irish Research Council. Outcomes to date include a collaboratively developed business plan and the co-design of the proposed integrated care pathway.

Comments on sustainability: Minimal initial funding is required (clinical care co-ordinator and some dedicated paediatrician sessions) because individual specialist services are in place. It is likely that there will be cost savings in the long term.

Comments on transferability: It is envisioned that this model could act as an exemplar for other rare complex disorders and improve support to community-based primary care teams in managing these conditions.

Keywords: 22q11.2 deletion syndrome; patient driven collaborative initiative; rare disease