

# Focus on Mainstream Health Care Access and Participation for Adults with Intellectual Disability due to Rare Causes

Robyn A Wallace\*

Department of Internal Medicine, University of Tasmania, Calvary Health Care, Tasmania, Australia

\*Corresponding author:

Robyn A Wallace

 robyn.wallace@utas.edu.au

Department of Internal Medicine, University of Tasmania, Calvary Health Care, Tasmania, Australia.

Tel: +61362262999

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## Abstract

At least half of adults with intellectual disability are estimated to have diagnosed or undiagnosed chromosomal or DNA mutations as the cause of their intellectual disability, which, by definition, are rare. Mostly however, associated physical health problems are not rare, and are well within the scope of mainstream health professional competencies. An unintended consequence of focus on the rareness of the cause of the intellectual disability, no matter how useful that is to understanding the syndrome, is that it deflects attention away from optimal care provision of the relatively common physical health problems that occur in this population, creates a barrier to inclusion into mainstream health access, and may contribute to the excessive preventable mortality and morbidity observed in this population. In this brief communication, a mainstream physician with experience in healthcare provision for adults with and without intellectual disability, a policy maker, researcher and educator in relation to healthcare for adults with intellectual disability offers a reasoned perspective that focusing on implementation of ways to optimize access to and participation in mainstream healthcare by adults with intellectual disability may lead to better quality and quantity of life for this population, instead of wholly focusing on development of separate services dedicated to exploration of the rareness of their cause of intellectual disability. In thinking about the place of rare syndrome organisations, the author proposes that within this mainstream model a small core of expert consultants who work within mainstream health who would oversee quality service, participate in development and implementation of reasonable adjustments within mainstream health settings and professionals, the health interface with the disability sector, support colleagues who are looking after adult patients with intellectual disability, and who are informed resources or researchers on rare syndromes of intellectual disability is consistent with the disability values of inclusion while also acknowledging the functional and biochemical aspects of syndrome of intellectual disability. The specialist skills of scientist and clinician experts and organisations in rare causes of intellectual disability in collaboration with mainstream clinicians, health services and the disability sector may contribute to better health outcomes with longer and better quality of life for adults living with rare causes of intellectual disability.

**Keywords:** Healthcare models; Adults with intellectual disability; Rare syndromes

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## Introduction

In Australia, a "rare disease" is defined as one with a prevalence of less than 5 in 10,000 people [1,2]. There are thought to be more than 7000 rare diseases, which as a group, affects up to 8% (or around two million) of Australians [3]. A National Strategic

Action Plan for Rare Diseases developed in 2020 to provide a comprehensive policy framework is based on three pillars: Awareness and Education, Care and Support, Research and Data [4]. Such frameworks highlight the special needs of people living with rare diseases and their families compared to people without

rare diseases, and appear to provoke a comfort of solidarity in uniqueness as expressed by a rare disease organisation motto “Alone we are rare” [5]. Improved professional training on rare diseases [6], enhancing patient active involvement as a source of information for clinicians [7], and identifying generic interventions targeting shared burdens among patients with rare diseases [8] are among the proposed recommendations to improve medical outcomes of people living with rare diseases. Adults with intellectual disability comprise a population for whom at least half of the causes of their intellectual disability is estimated to be due to an underlying chromosomal or DNA mutation [9], and which satisfy criterion for rareness. Extensive testing of etiology is a focus of specialist paediatrics systems especially given the importance of early intervention therapy, though diagnosis is still of value for adults [10]. The underlying mutation may impact upon known and unknown mechanisms in neuronal function, anatomy, metabolism, and as well also cellular functions in other organs, giving rise to the syndrome [11].

## Discussion

As a population, adults with intellectual disability have about 4 to 6 medical problems per person, variably associated with the rare syndrome, a lack of healthy living, iatrogenic causes, non-syndromal illnesses, and experience of negative social determinants of health [12]. Importantly, though the etiology of the intellectual disability may be rare, the types of medical problems in this population are ones frequently encountered by General Practitioners, or Specialist Consultants in any hospital in seeing adult patients without intellectual disability. Health professionals are well trained to manage and identify observed common problems within this population: risk factors for ill-health, polypharmacy, dental disease, sensory problems, epilepsy, thyroid disease, hypogonadism, gastroesophageal disease, *Helicobacter pylori*, constipation, osteoporosis and fractures, and accidents [12]. Despite this the diagnosis of health problems among adults with intellectual disability is often delayed and

**Table 1** Combining mainstream and specialist healthcare for adults with rare causes of intellectual disability.

1	Optimal availability and accessibility to mainstream health services and health professionals
	Requires development and establishments of reasonable adjustments to mainstream to facilitate access to and participation in healthcare by adults with intellectual disability: eg more time for consultations, adequate support for communication, adoption of a proactive approach to healthcare, sedation service for imaging, opportunistic testing
	No extra financial, physical, organisational or legislative barriers to use mainstream
	Participation in mainstream processes involving auditing of quality service, mortality and morbidity reviews, and other healthcare outcomes
	Adoption of a proactive approach to healthcare
	Aim for continuous improvement in service delivery and health outcomes
2	Organised access to core specialist group within mainstream
	Available for consultation by all health professionals (physicians, general practitioners, surgeons, psychiatrists, allied health professionals) with expertise in rare conditions of intellectual disability and having links with other related networks
	Maintains a direct clinical role in care of adults with intellectual disability with their families and disability support networks
	Provides information about rare disorders of intellectual disability in adults with appropriate levels of complexity to colleagues as well as adults with intellectual disability, their families and disability support networks. Information includes associated disabilities, medical issues, behavioural and cognitive phenotypes, treatments, and about living with intellectual disability.
	Provides clinical advice to colleagues about how to refine their biopsychosocial approach to taking a history, performing examinations, ordering tests, making diagnoses, management plans and review for their adult patients with rare cause of intellectual disability accommodating for impact of intellectual disability on communication and living circumstances and disability supports.
	Provide advice on testing for unknown cause of intellectual disability in adults
	Participate in review and oversight of quality of ethical aspects of health care
	Participate in overseeing framework in place for to ensure that adults with intellectual disability have adequate disability supports organizes to enable them to access and participate in mainstream health services
	Responsibility for improved education and competencies among mainstream colleagues on science, clinical aspects, and values in relation to healthcare of adults with intellectual disability within mainstream health settings
3	Organised and optimal disability supports plan in place to enable access to and participation in mainstream healthcare
	Requirement that adult patients with intellectual disability have established and robust disability supports plans in place which are activated when they need healthcare in any setting.
	Development of standards, expectations and specific roles of disability and family supports in healthcare settings
	Requires implicit understanding that disability supports be included, involved and accept responsibility in specialist core and mainstream healthcare with the individual adult patient with intellectual disability
4	Establishment of a health-disability interface within mainstream
	Involves adults with intellectual disability and their families as well as disability sector professionals to collaborate with specialist core and mainstream health professionals on types of reasonable adjustments, feedback on quality service aspects, service delivery and outcomes
	Facilitates cross fertilization of knowledge and values with respect to paradigms, policy, protocol, demarcation of duties, regulatory and legislative issues between disability and specialist and mainstream health sectors and professionals
	Clarifies practical responsibilities of health and disability sectors and professionals to adults with intellectual disability patients/clients in GP and specialist visits, public health and inpatient settings
	Carries information from collaboration into their own sector to increase knowledge and education about the other

not reaching recommended quality standards contributing to substantial preventable morbidity and earlier mortality among this population [13-15]. Otherwise well credentialed medical practitioners and health systems fail when it comes to providing optimal care to adult patients with intellectual disability, in part because the disability and its rare cause overwhelms and obscures the view of the patient as a person living with a rare disability with medical problems, in part because of barriers to access and participation imposed by health systems and professionals, and in part due to inadequate provision of disability supports in health settings [16]. In contrast to the case of patients with rare conditions not associated with cognitive impairment, adults with intellectual disability frequently have communication limitations and so are unable to independently be a source of helpful information to their health professional about their rare cause of disability. Furthermore, adults with intellectual disability may no longer have parents to facilitate sharing of information and disability supports may not have been included in previous discussions on etiology of disability. One can speculate that rare disease organisations with their focus on difference, isolation and disease inadvertently potentiate the view of seeing the patient as a "rare disease" as opposed to a person living with a rare disease with medical problems, thus alienating them and accidentally contributing to adverse outcomes. Furthermore, "intellectual disability" despite any rare etiology is not considered a disease but a "disorder" with onset in the developmental period with deficits in intellectual functioning and adaptive functioning [17]. Contemporary internationally accepted human rights disability values and systems promote the view that adults living with intellectual disability should have access to mainstream health services, rather than separate specialist intellectual disability services [18,19]. Success of this approach is acknowledged to require the presence of adequate disability supports for the individual with intellectual disability and of the development of reasonable adjustments to delivery of mainstream services from

health professional and health systems to optimize access to and participation in mainstream healthcare by adults with intellectual disability [20,21]. The benefits of specialist research and clinical care focusing on disease undertaken by rare disease facilities are undeniable as are the benefits of satisfying ethical human rights obligations of securing mainstream healthcare access for adults living with rare causes of intellectual disability. Conceivably both contribute to better quality and quantity of life lived by adults with intellectual disability. How can a healthcare service contain these seemingly incompatible elements for the good of adults with intellectual disability? A proposed solution is a framework incorporating a small core of specialised clinicians, services, and networks in rare causes of intellectual disability to back up and support (and not take over) mainstream equivalents, provision of adequate disability supports for adults with intellectual disability to enable access and participation in mainstream healthcare, development of reasonable adjustments to mainstream services, policies and processes, and establishment of a formal disability-health collaboration (also involving adults with intellectual disability and their families) within the health sector to enrich the service delivery (**Table 1**).

## Conclusion

In this proposed model, organisations of rare diseases have obvious important roles, such as those outlined in the Action Plan in contribution to the core specialist group work placed within mainstream. Their role is expanded to work with mainstream colleagues to oversee disability supports in place, design and implementation of reasonable adjustments and instigation of a disability-health interface. In these ways, the joint work of core specialist within larger mainstream accentuates the normality, the lack of unusualness, of having a rare condition in receiving best possible healthcare. It may be able to reassure people with rare diseases they are not so alone and rare after all.

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