New Zealand Paediatric Surveillance Unit

CHILDHOOD DEMENTIA IN AOTEAROA NEW ZEALAND

Summary

The childhood-onset dementias are an under-recognised and heterogeneous group of individually very rare disorders, with devastating impacts on affected tamariki, rangatahi and whānau. Research concerning childhood dementia (CD) has typically focussed on specific rare disorders, the majority of which remain untreatable. However, a collective definition has recently been proposed by the Childhood Dementia Initiative, to improve awareness, supports, and stimulate further research. In Australia and the UK, the estimated number of premature deaths per year caused by CD is equivalent to those caused by childhood cancer. The prevalence of CD in Aotearoa New Zealand has not been studied systemically, and thus the burden of disease is unknown.

Objectives

1. To estimate the collective prevalence of CD in Aotearoa New Zealand, with sub analysis for ethnicity and geographic distribution
2. To identify the aetiologies of CD in the NZ population
3. To determine the proportion of children with CD without a specific genetic diagnosis, with sub-analysis for ethnicity and geographic distribution
4. To determine the specialties involved in the care of CD patients, with sub-analysis for ethnicity and geographic distribution
5. To explore the diagnostic work-up, time from symptom onset to diagnosis and multisystem involvement in children diagnosed with CD in Aotearoa New Zealand

Case Definition and Reporting Instructions

Children with progressive neurocognitive decline, presenting before 18 years of age, as characterized by multiple losses of prior attained development skills in the context of generalised brain dysfunction, secondary to disease of suspected monogenic aetiology.

Inclusion criteria:

1. Sustained loss of developmental milestones in the context of generalized brain dysfunction
2. Skill loss most probably due to brain dysfunction.
3. Evidence of generalised (rather than focal) brain dysfunction.
4. Duration of illness greater than three months.
5. Suspected or confirmed monogenic (single gene) cause – even if this has not been confirmed
6. Presentation before 18 years of age
7. Patient does not need to be alive at the time of reporting

Examples of CD include, but are not limited to; adrenoleukodystrophy, Leigh syndrome, Batten disease, mucopolysaccharidoses, Huntington's disease

Exclusion criteria:

1. Regressive autism: non-sustained loss of developmental milestones limited to social/communication domains, considered to be consistent with a typical autistic regression
2. Acquired brain injury
3. Loss of developmental skills when brain dysfunction is not the primary aetiology

Please report any case of CD meeting the above inclusion and exclusion criteria.

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Follow-up of positive returns

A questionnaire requesting further details will be forwarded to practitioners who report a case.

Study background

In contrast to dementia in adults, childhood-onset dementia represents a heterogeneous group of disorders, which have collectively received minimal public recognition. Research concerning CD has typically focussed on specific disorders, such as Batten disease, Sanfilippo syndrome, X-linked adrenoleukodystrophy, Leigh disease, or other mitochondrial disease, and juvenile Huntington's disease. These individual disorders are very rare, typically affecting fewer than 1/50,000 children, and the majority remain untreatable.

The collective term 'Childhood Dementia' has recently been proposed, to improve awareness, supports, and stimulate further research. The Childhood Dementia Initiative was established for this purpose and has resulted in four publications and 12 ongoing research projects to date. This research has explored common symptoms and behavioural phenotypes, the psychosocial burden on families, and has informed collective approaches to care and development of treatments. Advocacy resulting from this initiative resulted in the inclusion of children in Scotland's Dementia policy, and the subsequent extension of dementia support services to children in 2023. It has also raised public awareness of CD and facilitated connection of affected families.

As part of this initiative, a scoping review estimated the burden of CD in Australia by pooling epidemiological data from the literature for individual disorders causing childhood dementia. This yielded a predicted incidence of 34.5/100,000 children, median survival of 9 years, and prevalence of 5.3/100,000 population. This was in concordance with a previous prevalence study conducted by the Australian Paediatric Surveillance Unit which yielded a prevalence estimate of 5.6/100,000. In addition, the authors emphasised that the estimated number of premature deaths per year caused by childhood dementia is equivalent to those caused by childhood cancer, and the comparative lack of attention that these disorders receive was highlighted.

The largest epidemiological study of childhood neurodegenerative disease performed to date, the Progressive Intellectual and Neurological Deterioration (PIND) study, has recently concluded in the UK. This British Paediatric Surveillance Unit study was created to identify cases of variant Creutzfeldt-Jakob disease but ultimately confirmed that the majority of diagnoses of CD were inherited metabolic disorders (IMD). This study found a similarly high incidence of IMD to studies in Australia but also identified significance ethnic disparities. The term PIND is not familiar or accessible to lay audiences; thus, the umbrella term CD is now preferred, and is gaining momentum amongst patient advocates, clinicians and researchers.

In addition to this prospective surveillance study, we plan to conduct a prevalence study of CD in Aotearoa New Zealand (NZHDEC approval pending).

Investigators

Rebecca Nicol,[1] Dr Isaac Bernhardt,[1] Dr Hannah Jones,[2] Dr Amanda Evans,[3] Dr Eloise Watson,[4] and Dr Emma Glamuzina.[1]
1 Adult and Paediatric National Metabolic Service, Starship Children's Health.
2 Paediatric Neurology, Starship Children's Health
3 Rei Kōtuku, Paediatric Palliative Care NZ
4 Neurology, Wellington Regional Hospital

New Zealand Paediatric Surveillance Unit Dunedin School of Medicine, P.O. Box 913, Dunedin Tel: 03 556 6024 Email: nzpsu@otago.ac.nz

THANK YOU FOR YOUR HELP AND SUPPORT THE RESULTS OF THIS SURVEILLANCE WILL BE INCLUDED IN THE ANNUAL REPORT OF THE NZPSU WHICH WILL BE AVAILABLE ON THE NZPSU WEBSITE AND CAN BE REQUESTED DIRECTLY FROM NZPSU

References

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