murdoch children's research institute

Australian Hand Difference Register

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Annual Report 2024

Foreword

The Australian Hand Difference Register (AHDR) is a database of children born with a congenital upper limb difference (CULD).

Children born with a congenital hand difference are a diverse group who, with their families, experience a wide range of functional and social impacts.

While hand differences and their associated treatments have been thoroughly described and classified, the data that inform families and assist in decision-making are sparse and often historical.

It is this gap that the Australian Hand Difference Register (AHDR) aims to fill. We provide nationally collected and standardised data on children with CULD, including classification, family demographics and long-term outcomes to assist patients, families and treating clinicians.

Established in 2017, the AHDR monitors the numbers of children diagnosed with a hand/arm difference across Australia and maintains a data collection of clinical and sociodemographic characteristics. The purpose of the register is to:

- generate evidence to improve clinical care and health service design
- understand the lived experience of CULD for children and families
- improve our understanding of causes and risk factors
- build genuine research partnerships with families to answer questions that matter most to them

News

Grant Success

A/Prof David McCombe received a research grant from the Australian Society of Plastic Surgeons. The $10,000 contributed to the 'Appearance Study: A comparison of tools used to assess hand appearance in children with congenital upper limb differences (CULD)'.

Participant Newsletters

A bi-annual e-newsletter was launched and sent to AHDR participants. Newsletters include relevant news and updates about the AHDR, information on current and upcoming research opportunities, relevant resources and reminders. The newsletters are designed to keep participants informed, engaged and connected to the register.

Giving Page

A 'Giving Page' was created for the AHDR, which is hosted on the MCRI website. The page summarises the AHDR's purpose and activities, and more importantly provides an opportunity for individuals to donate to support ongoing research projects.

To support the 'Giving Page' on the website, a two-page flyer was also created and distributed to the AHDR clinics.

New Site Opened

HNEKidsRehab was welcomed as a new recruitment site in Newcastle, New South Wales. Leslie Wollin and the team joined the AHDR in November 2024.

Current Research Projects

In 2024, new research projects were started to further explore CULDs from multiple perspectives. Through a combination of qualitative and quantitative approaches, the projects examine a range of clinical, social and personal factors. Together, they aim to generate new insights into clinical outcomes, lived experience and inclusion for individuals with CULD.

- A Comparison of Tools used to Assess Hand Appearance in Children with Congenital Upper Limb Differences (CULD)
- The Impact of Socioeconomic Status on Congenital Upper Limb Difference Registry Inclusion – Data from the Australian Hand Difference Register
- Analysis of Australian Experience with Radial Polydactyly - classification, treatment and patient rated outcomes
- Adolescent and Parental Perspectives of Living with a Congenital Upper Limb Difference - a qualitative study

Publications

Wilks D, McCombe D. *Developing a Congenital Upper Limb Difference Registry in Australia*. J Hand Surg Asian Pac Vol. 2024;29(6):486-491. doi:10.1142/S2424835524300068

This paper describes the development of the AHDR, including its structure, purpose and the challenges of capturing meaningful, standardised data across a diverse patient population. It highlights the importance of the register and how it can support more consistent data collection and national collaboration to improve care for children with CULDs.

Presentations

Combined Meeting of The American Society for Surgery of The Hand (ASSH) and The Australian Hand Surgery Society (AHSS), Hawaii, May 2024
An Analysis of the Functional and Psychosocial Impact of Congenital Upper Limb Differences Using PROMIS®: Data from the Australian Hand Difference Register – Dr Daniel Wilks

NSW Paediatric Rehabilitation Services (PRS) Education Day, August 2024
Australian Hand Difference Register – Ms Joanne Kennedy

Asia Pacific Federation of Societies for Surgery of the Hand (APFSSH) & Federation of European Societies for Surgery of the Hand (FESSH) Academy Foundation Course, Kuala Lumpur, August 2024
Longitudinal deficiencies and Phocomelia – A/Prof David McCombe
Talking to Parents about Congenital Upper Limb Differences (CULDs) – A/Prof David McCombe

Indian Society for Surgery of the Hand (ISSHCON) Annual Meeting, Bengaluru, October 2024
Talking to Children and Parents about CULDS – A/Prof David McCombe

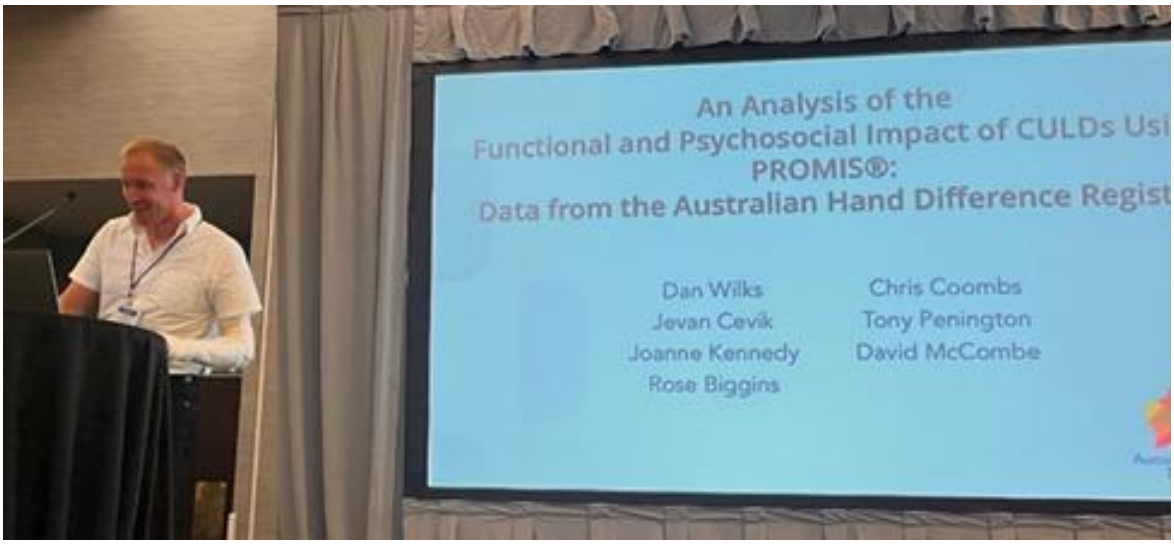

Photo 1. Dr Daniel Wilks presenting at the ASSH & AHSS combined meeting in Hawaii, May 2024

Data in 2024

In 2024, a total of **196 new notifications** were recorded, representing an increase from the 160 notifications reported in 2023. There have now been **1,618 children registered** on the AHDR since 2017. In addition, a minimum dataset is available for 1,142 children who were in the database before the AHDR was implemented. We therefore have a total of 2,760 individuals known to the AHDR.

Notifications were higher among males (n = 128) than females (n = 68). This higher percentage (65%) of male notifications is greater than in previous years, when it has been an average of 52%.

The median age of participants was one year, with over half of the notifications relating to children aged between one and three years. **Figure 1.** illustrates the percentage breakdown of the participants across the different age groups.

Place of birth was most commonly Victoria (38%), followed by Queensland (27%). Other states and territories contributed small proportions. **Figure 2.** describes the distribution of notifications in 2024 by place of birth.

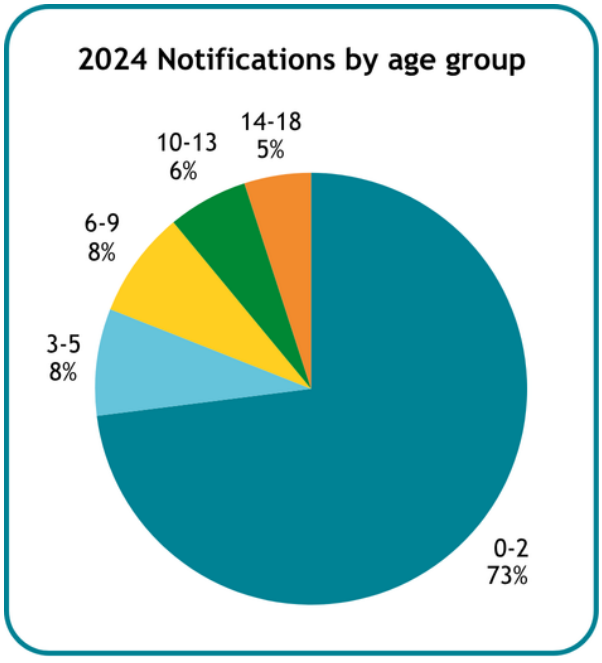

Figure 1. Percentage of notifications by age group (years) in 2024

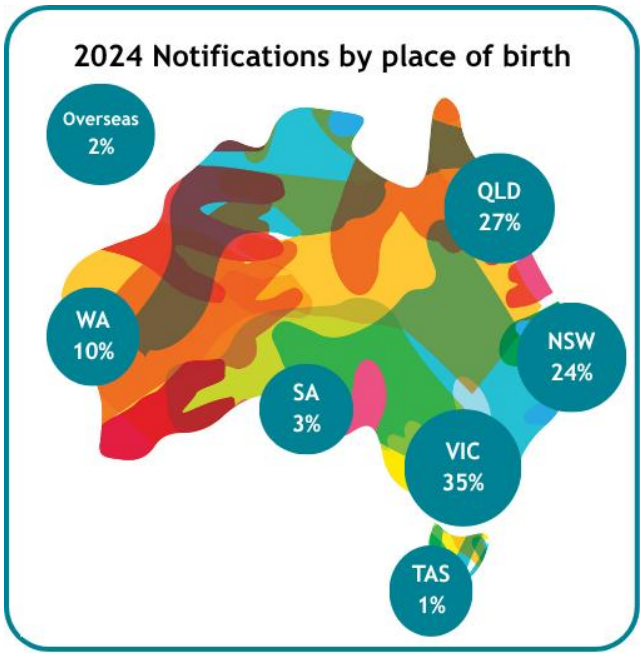

Figure 2. Percentage distribution of notifications by place of birth in 2024

The Oberg-Manske-Tonkin (OMT) classification is a system used to categorise congenital upper limb differences based on embryological development. It currently comprises 119 conditions.

The AHDR has collected data on 79 of these conditions, encompassing both primary and secondary diagnoses. In 2024, data was collected on 39 primary and secondary diagnoses. Table 1 presents the top five diagnosed conditions in 2024, with **radial polydactyly** remaining the most common diagnosis at **17%**. This is consistent with trends observed in previous years. Read below for a spotlight on radial polydactyly.

	Top 5 conditions in 2024	%
1	Radial polydactyly	17
2	Ulnar polydactyly	12
3	Symbrachydactyly (with ectodermal elements)	9
4	Soft tissue - cutaneous (simple) syndactyly	7
5	Isolated campodactyly	7

Table 1. Top 5 diagnosed conditions in 2024, presented as a percentage of total cases (n=196)

Questionnaires

At enrolment, **a one-off questionnaire** is sent to participants who have the option to complete it. It can be completed by the mother, father, guardian, or a mature minor. The questionnaire collects information on pregnancy, family history and the child's health. In 2024, the AHDR received **85 completed questionnaires**, a response rate of 43% from the new registrants. The majority were completed by mothers (81%), followed by fathers (18%) and guardians (1%).

At the ages of 5, 8, 11, 14 and 17 years, the Patient-Reported Outcomes Measurement Information System (PROMIS) questionnaires are sent to participants. The PROMIS measures assess key areas of health and wellbeing, including upper-extremity function, peer relations, depressive symptoms, anxiety symptoms, global health and pain interference. These insights contribute valuable information on the lived experiences of children with upper limb differences at various developmental stages.

In total, the AHDR has sent out 391 PROMIS questionnaires and collected patient-reported outcome data from **215 participants** across the age groups, a response rate of 55% from participants. The highest number of responses came from youngest age group - children aged 5 (n=72). The number of responses decrease with age, as illustrated in **Figure 3**. An analysis is currently underway to examine the functional and psychosocial impacts of CULDs during the first two years of data collection.

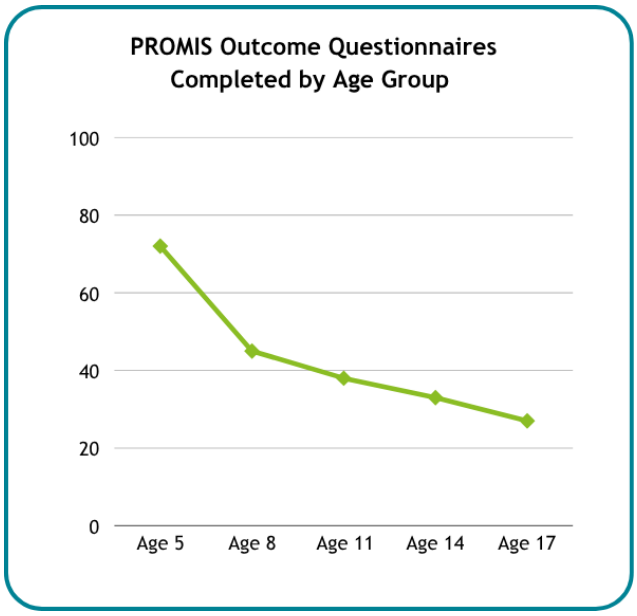

Figure 3. Total number of AHDR participants that have completed PROMIS Outcome Questionaires by Age

Diagnosis Spotlight: Radial Polydactyly

Radial polydactyly, also known as thumb duplication, is a congenital hand difference seen in approximately 0.08-1.4 per 1000 live births. It may occur sporadically, be inherited or present as part of a syndrome.

This following spotlight explores radial polydactyly as both a primary and secondary diagnosis. The analysis includes diagnoses recorded prior to the establishment of the AHDR, offering insight into the overall context of this condition.

In the AHDR, a total of **440 diagnoses** of radial polydactyly have been recorded, including 149 pre-AHDR entries and 291 notifications since the register began.

Over the past five years, a total of 188 diagnoses of radial polydactyly have been reported to the register. **Figure 4.** illustrates the proportion of radial polydactyly diagnoses (13-22%) in comparison to other conditions recorded in the AHDR.

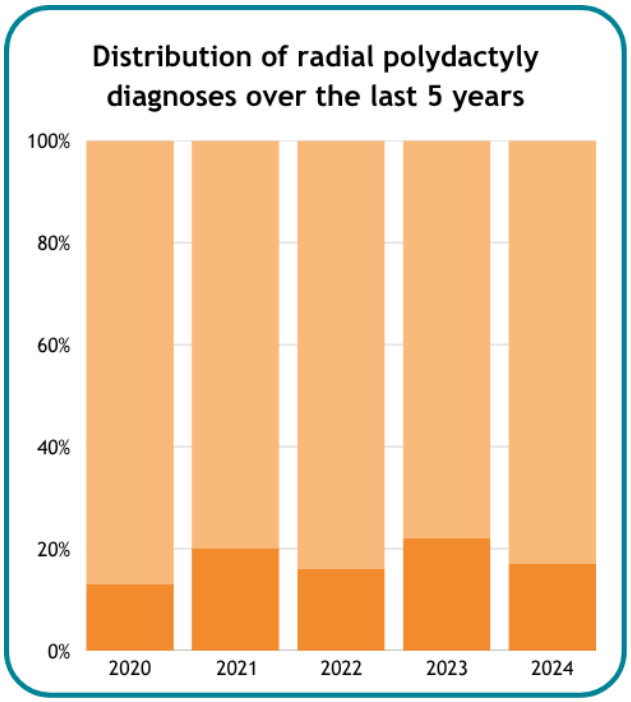

Figure 4. Percentage of radial polydactyly diagnoses recorded in the AHDR each year from 2020 to2024.

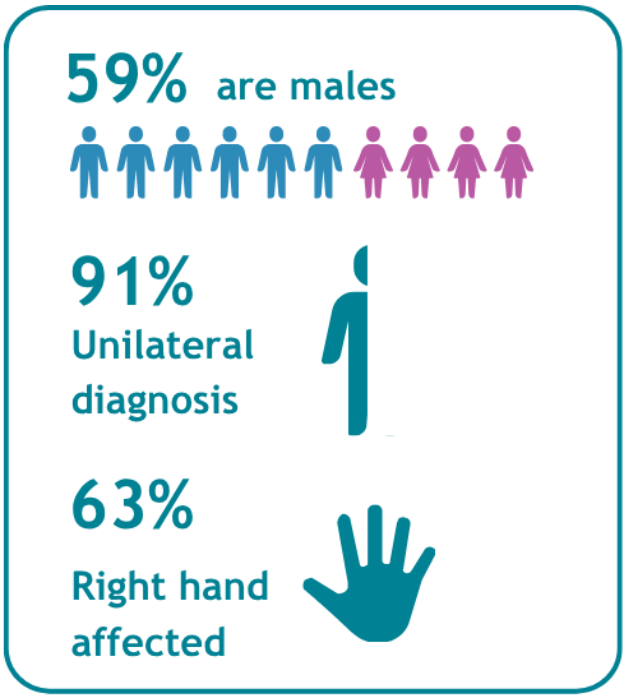

Figure 5. Summary profile of AHDR radial polydactyly participants diagnosed with radial polydacyly, highlighting gender and laterality

In the AHDR population, there are slightly more males than females diagnosed with radial polydactyly, with **males representing 59%** of all cases recorded.

The condition is predominately unilateral, with over 90% of cases only involving one hand. Of these, unilateral cases, the **right hand is more commonly affected (63%)** than the left (**Figure 5**).

Diagnosis occurs primarily at birth, with 91% of cases identified at birth. Conversely, prenatal diagnosis remains relatively uncommon, representing only 9% of cases (**Figure 6**).

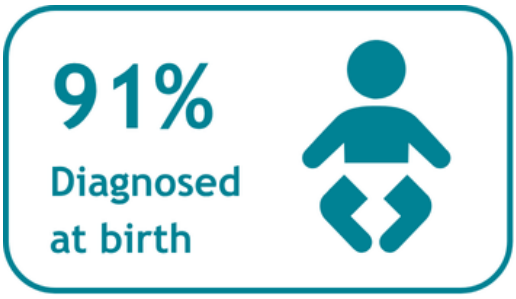

Figure 6. Percentage of radial polydactyly participants diagnosed at birth

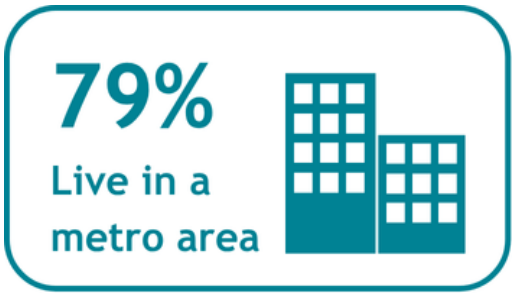

Figure 7. Percentage of participants residing in metropolitan areas based on available postcode data.

Of the 440 reported cases of radial polydactyly in the AHDR, postcode data was available for 223 participants. Of these participants **79% live in a major city or metropolitan area** and 21% live rurally (**Figure 7**).

Among children with radial polydactyly, 94% do not have a biological parent with a hand difference **(Figure 8).**

Additionally, 77% report no known hand differences among extended family members or relatives (**Figure 9**). These findings suggest that non-familial factors may play a role in the occurrence of radial polydactyly.

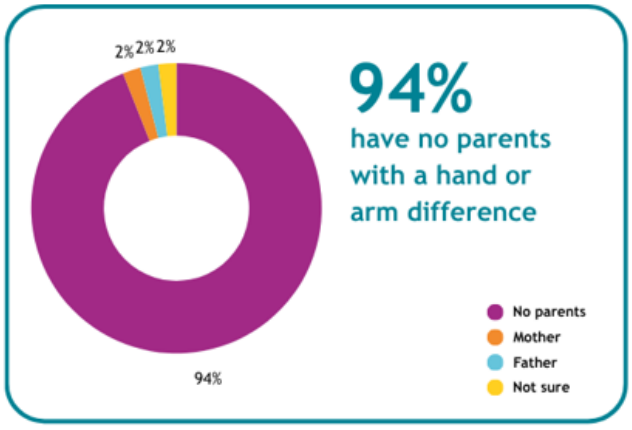

Figure 8. Percentage distribution of parents of participants with a hand or arm difference

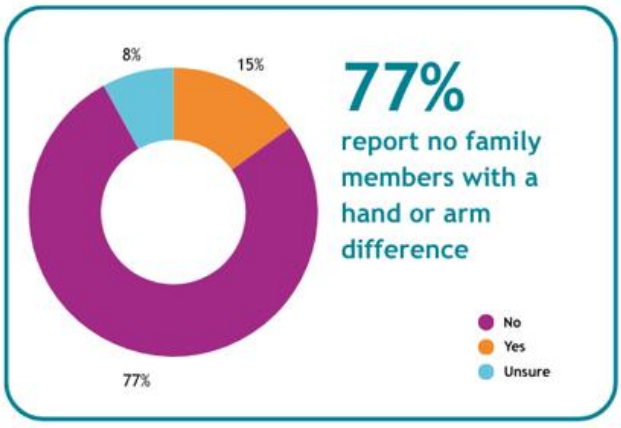

Figure 9. Percentage distribution of family members of participants with a hand or arm differnce

Research is currently underway to further understand radial polydactyly using data from the AHDR. It will explore the demographics and clinical characteristics in the Australian pediatric population. This information will be available in upcoming publications.

Summary

Over the past year, the AHDR has made steady and meaningful progress toward our goals. Our research has strengthened understanding in key areas and laid important groundwork for future activities. We acknowledge the valuable contributions of our participants, team, and supporters throughout this period. Looking ahead, we remain committed to building on this progress and ensuring the ongoing relevance and impact of our work.

Acknowledgments

Team

Joanne Kennedy (AHDR Manager)

Olivia Gigli (Research Assistant)

Steering Committee

A/Prof David McCombe

Dr Daniel Wilks

Prof Tony Penington

Dr Richard Lawson

Ms Rose Biggins

Ms Joanne Kennedy

Ms Olivia Gigli

Ms Elizabeth Borg

Recruitment Sites

Adelaide Plastic & Hand Surgery (SA)

Adelaide Women's & Children's Hospital (SA)

Flinders Medical Centre (SA)

Gold Coast University Hospital (QLD)

Hunter New England Kids Rehab (NSW)

Monash Children's Hospital (VIC)

Mudgeeraba Hand Clinic (QLD)

North Shore Private Hospital (NSW)

Perth Children's Hospital (WA)

Queensland Children's Hospital (QLD)

The Royal Children's Hospital (VIC)

Royal Hobart Hospital (TAS)

Royal North Shore Hospital (NSW)

Southern Plastic Surgery (VIC)

Sunshine Hospital (VIC)

Sydney Children's Hospital, Randwick (NSW)

The Children's Hospital, Westmead (NSW)

Victorian Clinical Genetics Services (VIC)

Victorian Hand Surgery Associates (VIC)

Western Australian Plastic Surgery Centre (WA)

Clinician's private rooms (national)

Funding

McNally Family Foundation

Australian Society of Plastic Surgeons

Thankyou

The AHDR team would like to thank the families who have generously shared their time and experience by participating in the register. We extend our heartfelt thanks to the McNally Family Foundation for their generous and ongoing support in funding Australian Hand Difference Register. We are also deeply grateful to all donors whose contributions help make this vital work possible. Your support is advancing research and making a real difference to the lives of children and families with hand differences. We would also like to thank Aussie Hands for their ongoing support.

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