Ethical implications and social acceptability of

X-ALD SCREENING IN FEMALE NEWBORNS

Australia's newborn bloodspot screening (NBS) programs identify newborn babies at risk of developing a serious condition.

Adding new conditions to NBS programs follows rigorous assessment via the decision-making pathway. This involves evidence reviews, health technology assessments, expert input, and consideration of alignment to the NBS National Policy Framework criteria. As part of this assessment process, advice is provided by the Medical Services Advisory Committee (MSAC) to support Australian Health Ministers to consider conditions for screening.

The purpose of this document is to present a concise summary of the work undertaken examining the ethical considerations and social acceptability of screening X-linked adrenoleukodystrophy (X-ALD) in female newborns.

ABOUT X-ALD

X-ALD is a rare genetic condition resulting from pathogenic variants in the *ABCD1* gene on the X-chromosome. X-ALD has three types of clinical manifestations with different levels of severity, symptoms, and age of onset. As a X-linked disease, the condition affects males and females differently.

MALES are **more likely** to be **affected by** X-ALD as they have one X-chromosome and one Y-chromosome.

FEMALES have two X-chromosomes, so their unaffected X-chromosome can help to **counter the affected X-chromosome**.

WHY WAS THIS WORK CONDUCTED?

MSAC provided advice that supported the inclusion of X-ALD in Australian NBS programs for all newborns. Considering this, in late 2024, Australian Health Ministers agreed to add X-ALD to Australia's NBS programs for male newborns, requesting further examination of the ethical considerations and social acceptability of screening female newborns.

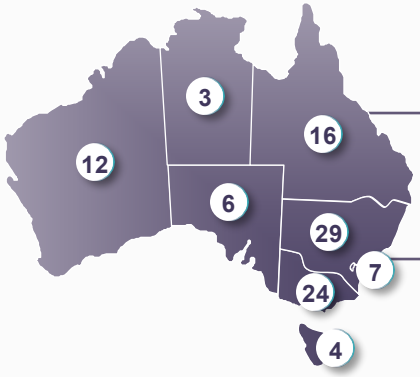
Following completion of this work, on 12 December 2025, Ministers agreed to also screen female babies for X-ALD within NBS programs. All governments have agreed that screening will begin within 2 years of health ministers' decision to add a condition.

METHODOLOGY

The work to explore ethical considerations and social acceptability of screening for X-ALD in female newborns involved consultations across Australia and an evidence review:

- **101** Members of the **general public** consulted through focus groups
- Interviews with **people with lived experience** of X-ALD **24**
- **17** Interviews with **expert stakeholders**
- **REVIEW OF INTERNATIONAL PEER REVIEWED LITERATURE**

GENERAL PUBLIC CONSULTATION

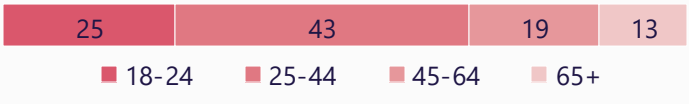


15 Focus groups

Female 55%

Male 45%

Age distribution (%)



Each theme in the summary may encompass a mix of benefits, drawbacks, and considerations, reflecting the complexity and nuance of the perspectives presented.

GENERAL PUBLIC

The majority of participants were **in favour of** the inclusion of **screening** for X-ALD in **female newborns**

Key themes raised

- **Potential informed future reproductive decisions** and **lifestyle planning** for the female with X-ALD and her family
- **Equality** for females was commonly expressed as an important reason to include females in screening
- **Potential psychological impact** to the female newborn's parents and the female when she learns of the result
- **Potential impact** on the **child-parent relationship**, such as heightened vigilance potentially impacting a child's upbringing

In a short poll:

of participants said **'Yes'** that screening for X-ALD in female newborns should be included in NBS programs

of participants said **'No'** to screening for X-ALD in female newborns, while **8%** were not sure or felt they did not have enough information

PEOPLE WITH LIVED EXPERIENCE

All participants with lived experience of X-ALD **supported** the inclusion of **screening** for X-ALD in **female newborns**

Key themes raised

- Experiences of **missed** or **misdiagnosis of X-ALD** to highlight the benefits of newborn screening for females
- Having **future, informed reproductive and lifestyle planning** and **cascade testing**
- Receiving **an earlier diagnosis** and access to timely **symptom management**
- Ensuring **equality in access** to information, healthcare and symptom management
- **Potential psychological impact** of knowledge of X-ALD status on the female newborn and family

All factors can impact overall **quality of life**

EXPERT STAKEHOLDERS

There were mixed perceptions about the potential screening for X-ALD in female newborns.

Key themes raised

- Many expert stakeholders agreed on the **secondary benefits** of screening in females (e.g. future, informed reproductive decisions for the female and family)
- However, experts held mixed perceptions on the **weight** of such benefits, in the **absence of a direct benefit** to the female newborn
- Some experts perceived that the **established national policy framework criteria do** not indicate screening for X-ALD in female newborns should be included
- Most discussions focused on the considerations for potential screening, including **practical** and **health system implications**

LITERATURE REVIEW

Evidence reviewed suggests that there is **no global consensus** on acceptability of screening for X-ALD in female newborns, and a range of ethical considerations.

There is **limited literature** that discusses ethical issues and psycho-social outcomes of screening females for X-ALD. N**o global consensus exists** on its acceptability with countries including varied screening protocols. **International screening criteria do not provide a clear answer** for X-ALD in the Australian context.

- The main justification for screening females is to enable **cascade testing** to identify at-risk male relatives. Other justifications include **informing future reproductive decisions**, **raising awareness** of X-ALD and **avoiding diagnostic delay**.
- The main concern with screening females for X-ALD is that there is **no direct benefit to the female newborn**. Other drawbacks include **living under uncertainty**, overriding the right not to know, **resource implications**[1] from follow-up counselling and care, potential insurance discrimination, **setting a precedent** for screening other X-linked conditions and a possible **loss of public trust** in screening.
- Parents and females with lived experience generally support having information via NBS or later in life, such as during adolescence or as part of carrier screening.
- Counselling and clinical follow up may be more complex in screen-positive female newborns than it is for male screen-positive newborns.

[1] *In September 2024, the Assistant Treasurer announced a total ban on the use of adverse genetic test results by life insurers. This was introduced into Parliament November 2025 and will need to be implemented via legislative changes.*