

Population approaches to the primary prevention of birth defects

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Acknowledgements

- Research collaborators
- Consumer and community advisors and representatives
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- Funders – government and non-government



Overview of presentation

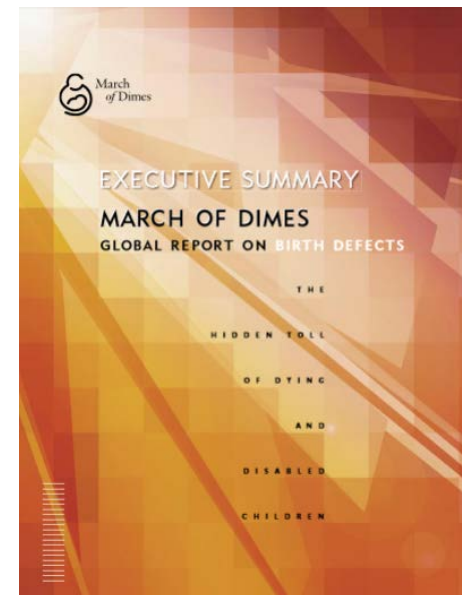
- Why are birth defects important for early childhood development?
- Primary prevention:
 - Evidence, engagement and evaluation
 - Two case studies:
 - Neural tube defects (NTD)
 - Fetal Alcohol Spectrum Disorder (FASD)
- Conclusions



Why are birth defects important?

- 7.9 million children—6% total births worldwide
- ~3.3 million < 5 years die from birth defects each year
- ~ 3.2 million of those who survive have disability
- Global problem, impact is particularly severe in middle- and low-income countries

March of Dimes Global Report on Birth Defects. Christianson et al, 2006



Why are birth defects important?

- Birth defects - major cause of mortality
- Birth defects - major cause of morbidity
- Causes unknown for many
- Some amenable to primary prevention
 - Many to treatment
 - Few to cure
- Affect early development



Population approaches to primary prevention— evidence, engagement, evaluation



3 E's: EVIDENCE – ENGAGEMENT - EVALUATION



Neural tube defects



Neural tube defects

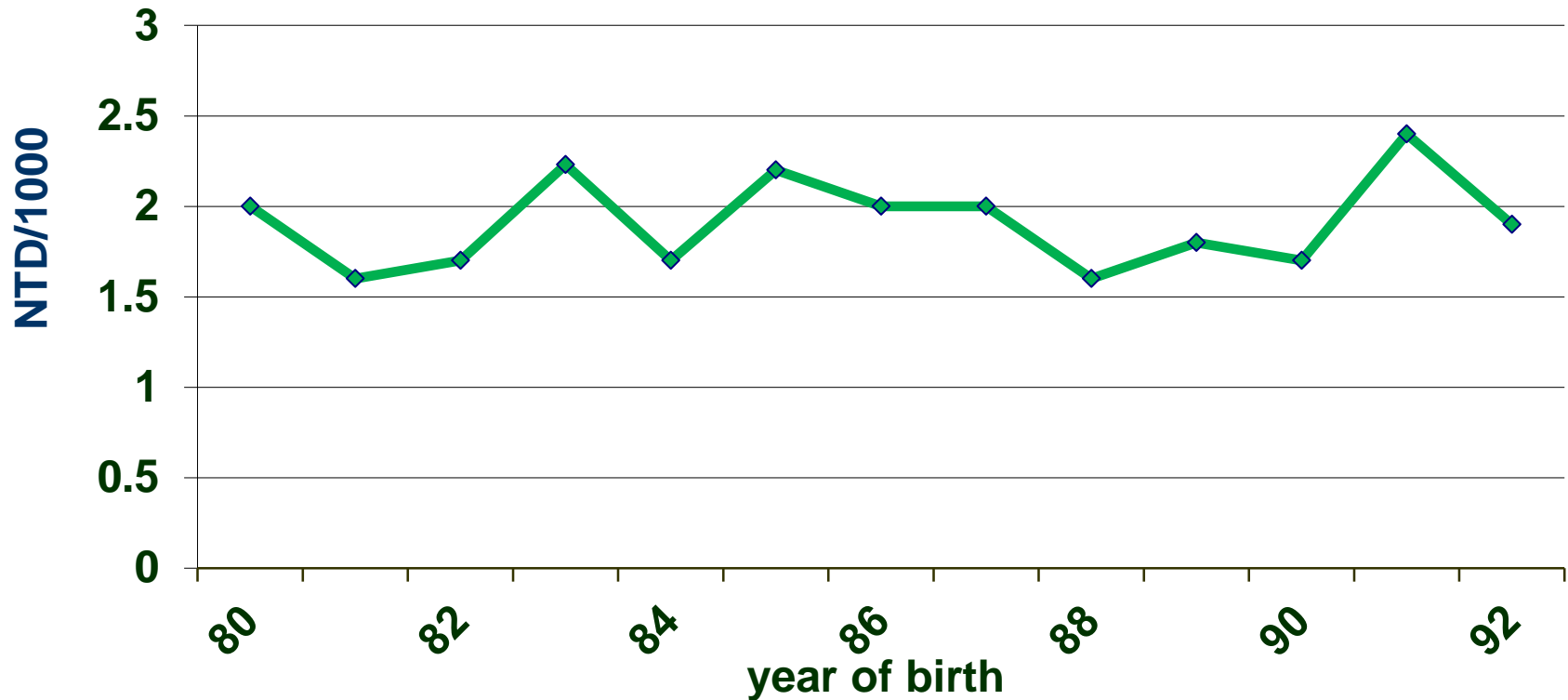


Spina bifida



Anencephaly

Neural tube defects Western Australia



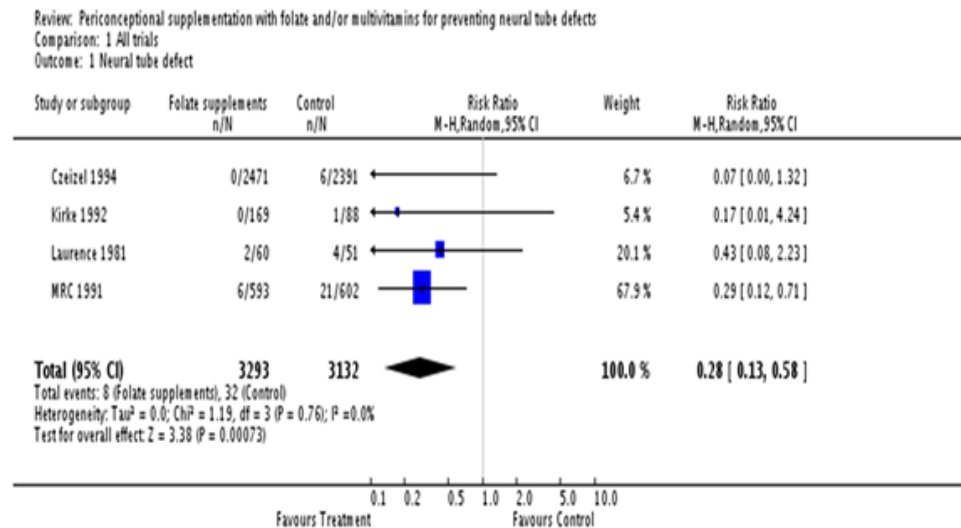
NTD 40% more common in Aboriginal vs non-Aboriginal infants

Bower et al 1989

Evidence of effect of folic acid

- Many observational studies showing reduction in NTD with increasing folate intake

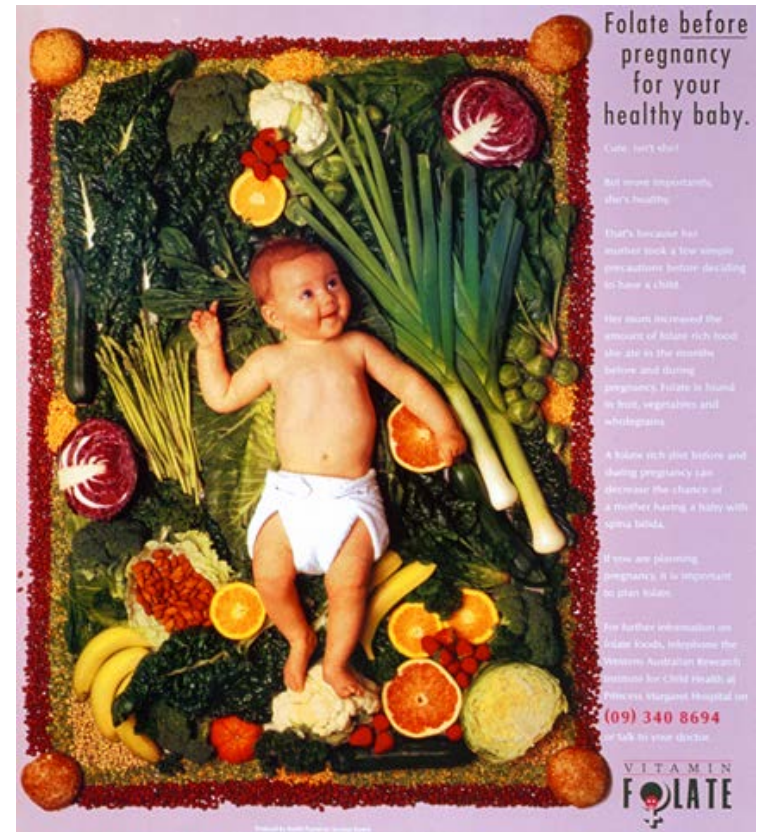
- 1992 - confirmed that peri-conceptual **folic acid** supplementation prevents 70% NTD



Health promotion program 1992+

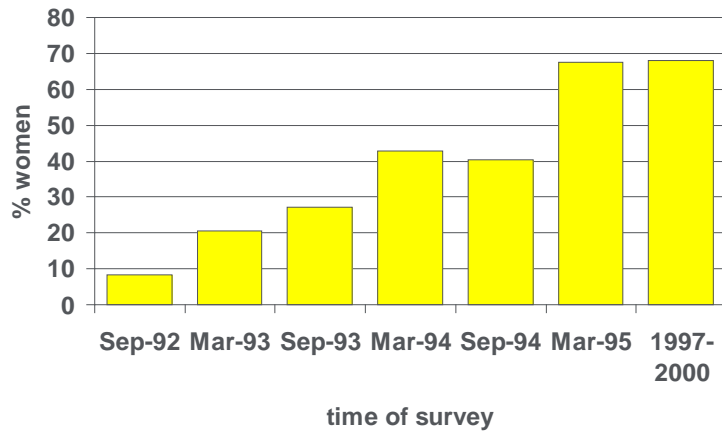
Engagement with consumers and stakeholders

- Inform health professionals of folate-NTD link
- Inform women of childbearing age
- Promote use of folic acid supplements
- 1996 voluntary fortification of some foods

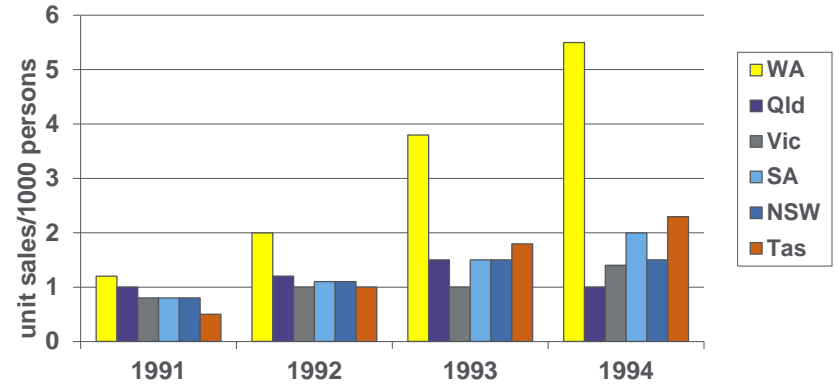


Evaluation of interventions

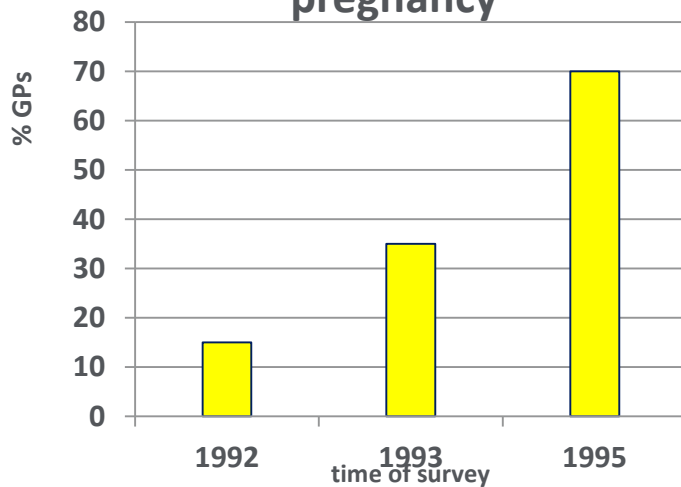
Women's knowledge of link between folate and spina bifida



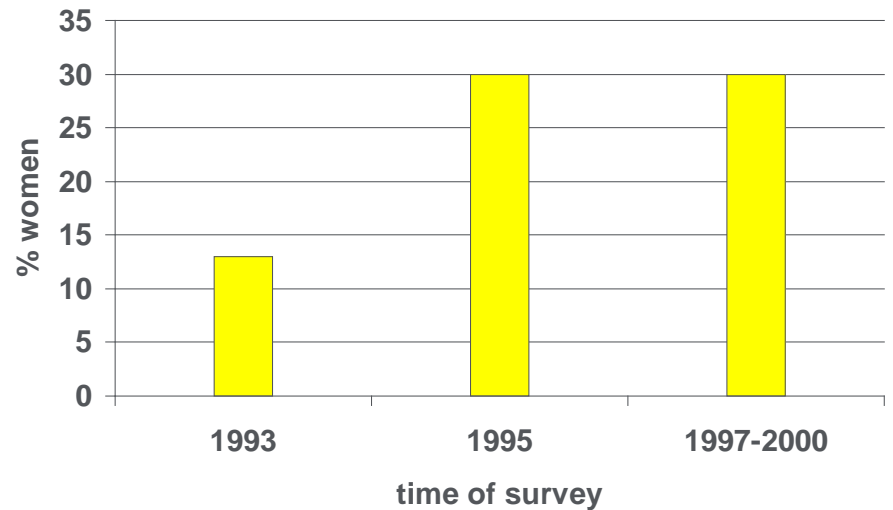
Sales of folic acid supplements



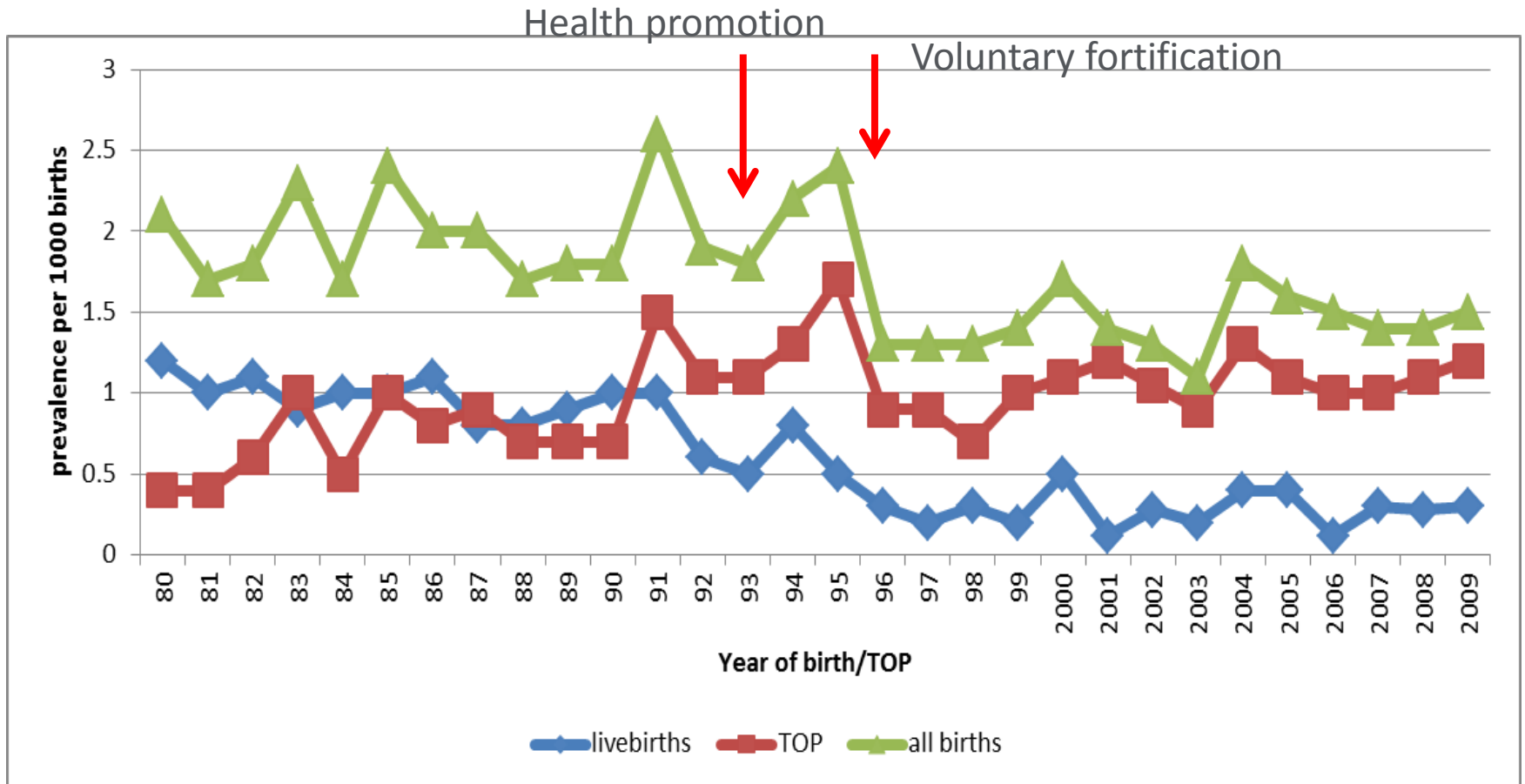
Proportion of WA GPs advising folic acid supplements to women planning a pregnancy



Proportion of WA women taking folic acid supplements periconceptionally

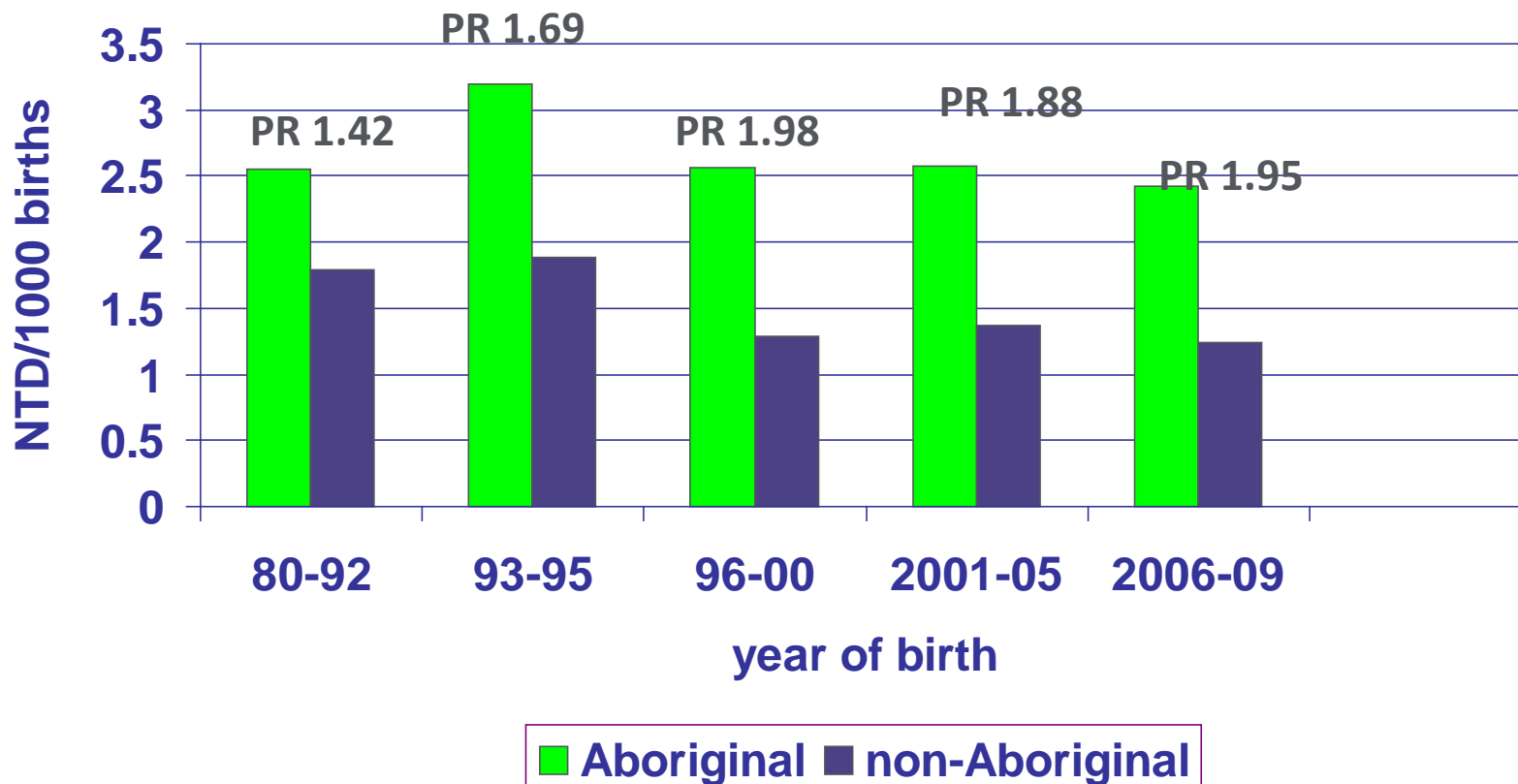


Neural tube defects, Western Australia



But no change in high rate amongst Aboriginal infants

NTD in Aboriginal and non-Aboriginal infants by grouped years of birth





Evidence, evaluation and engagement

Health promotion, voluntary fortification:

- Not maximally effective - only ~30% reduction in NTD
- Few foods voluntarily fortified, mainly breakfast cereals
- At best, 40% women take folic acid periconceptionally
 - Inequities in folic acid supplement use - Young women, smokers, less well-educated, unplanned pregnancies
 - Folate must be taken BEFORE and in early pregnancy
 - Unplanned pregnancies – 40-50%
 - Continual education
- not working for Aboriginal people and gap widening



consideration of mandatory fortification



Evidence, evaluation and engagement

Renewed Engagement

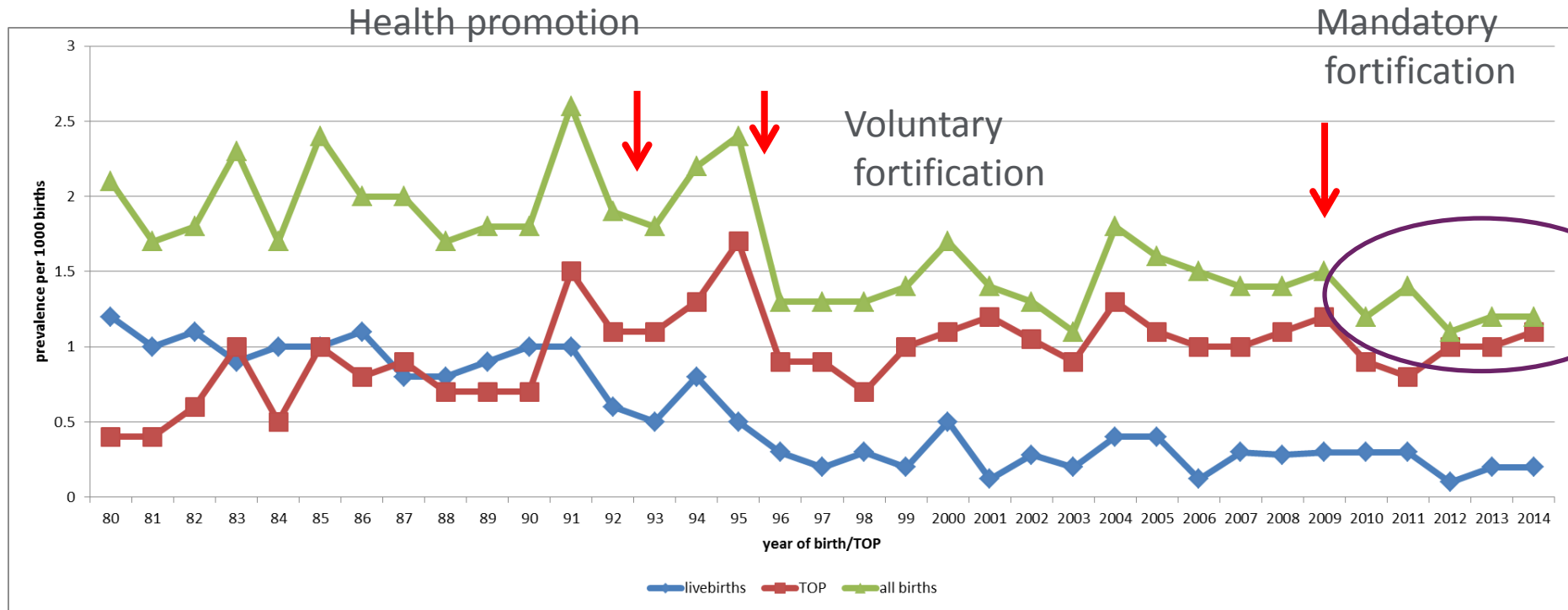
- Spina Bifida Association
- Departments of Health
- Food Standards Australia New Zealand
- Health professionals
- Aboriginal community, researchers and leaders

Evidence from this research, in conjunction with other evidence, led to:

Mandatory fortification of wheat flour for bread-making with folic acid in Australia in 2009

Evaluation of mandatory fortification

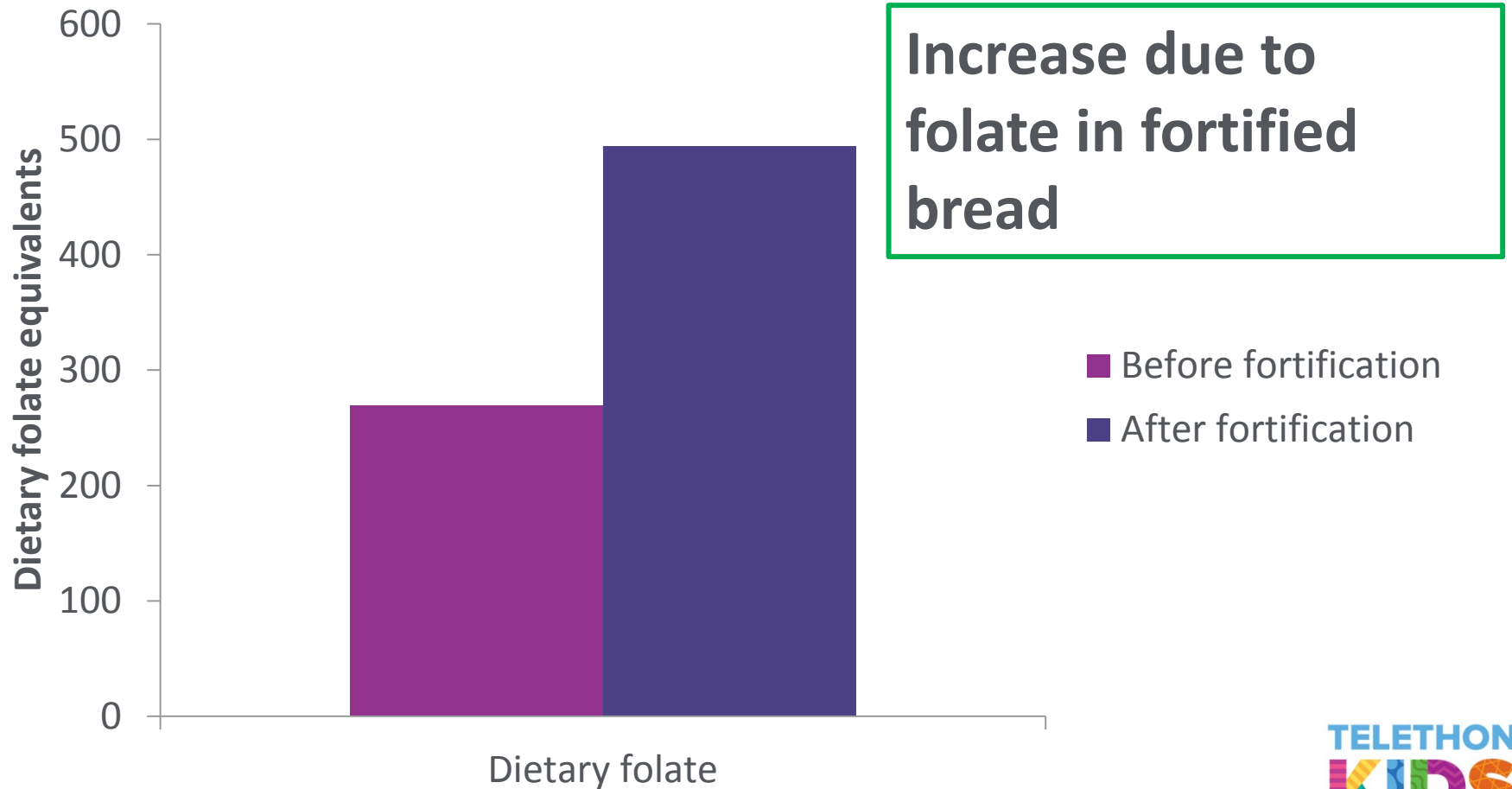
Neural tube defects, Western Australia




Further 15% reduction in NTD since 2009



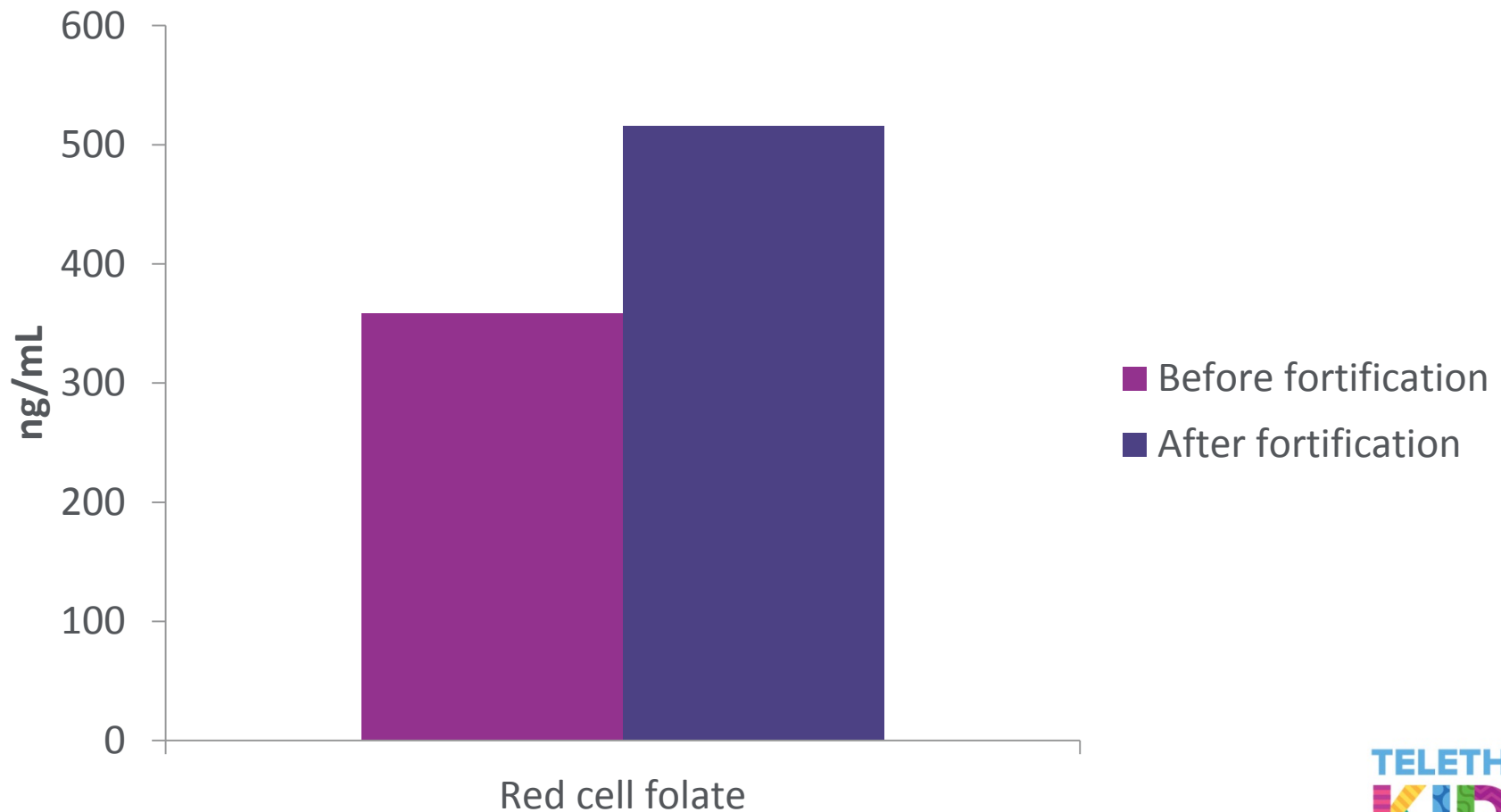
Mean dietary folate intake, Aboriginal people, Western Australia



Bower et al, 2015

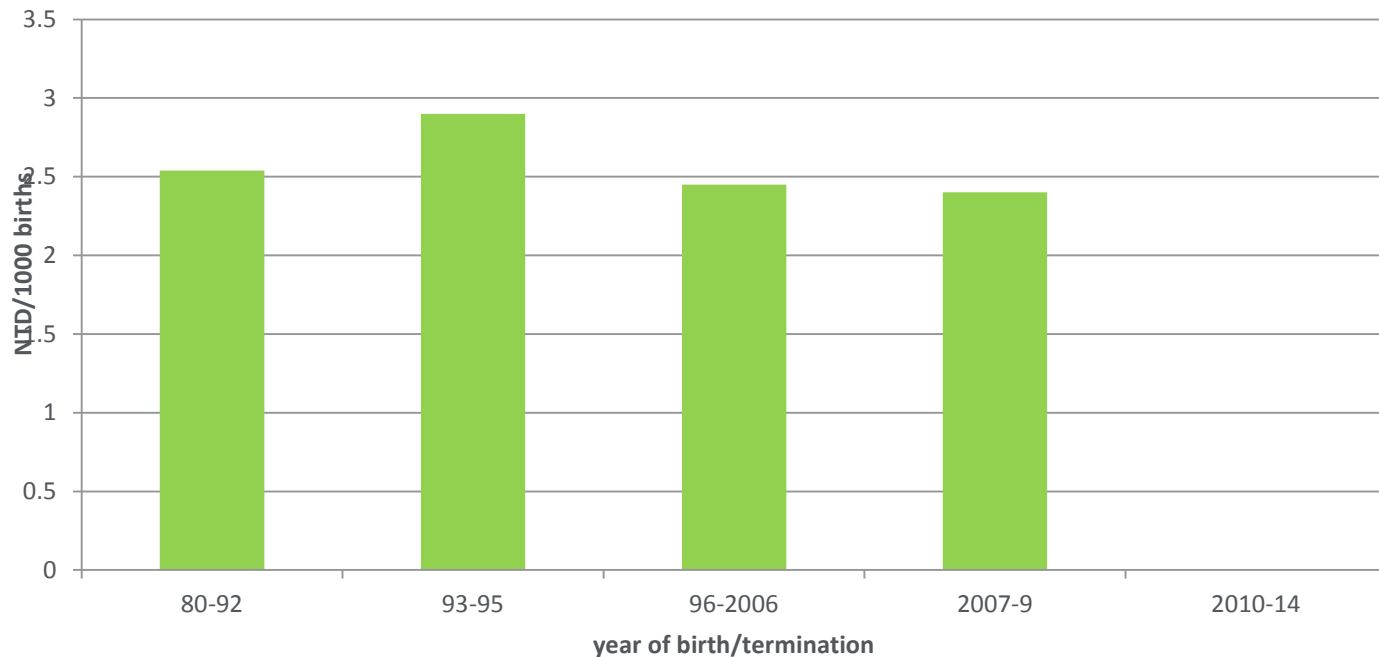


Mean red cell folate, Aboriginal people, Western Australia



Bower et al, 2015

Neural tube defects, Aboriginal infants, Western Australia



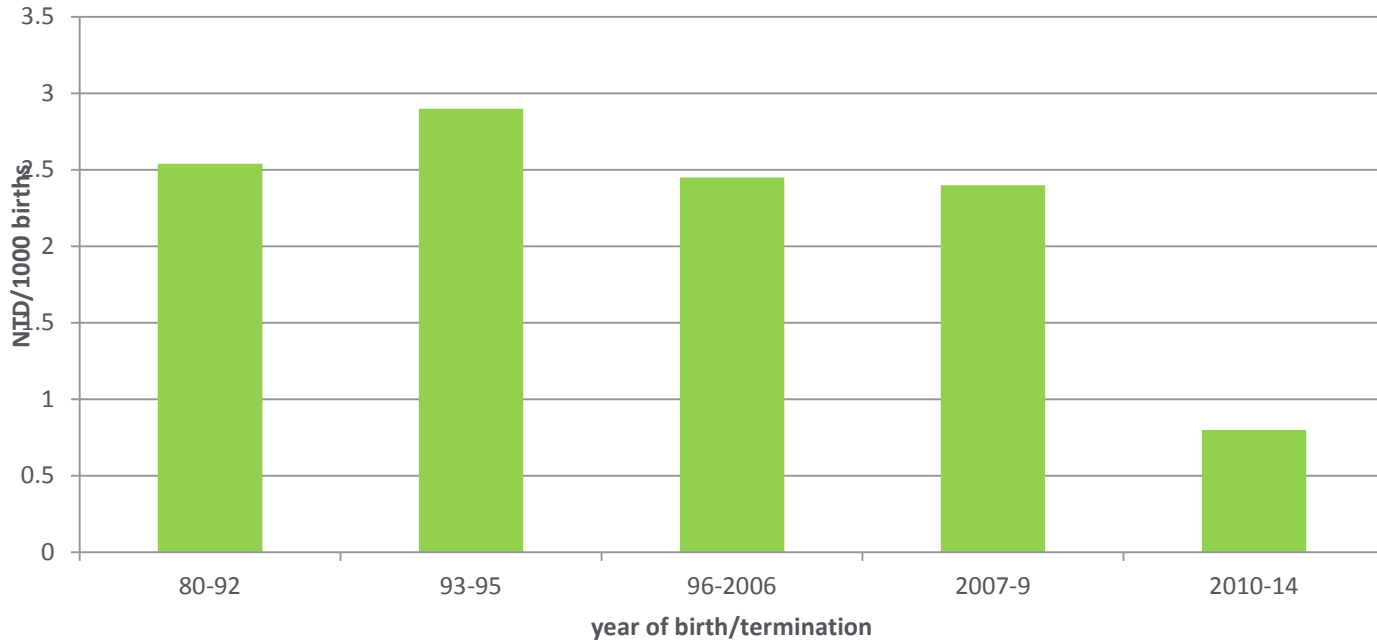
1980-92 –before any health promotion or fortification

1993-95 –health promotion of folic acid supplement use

1996-06 –health promotion & voluntary fortification

2007-09 –transition period

Neural tube defects, Aboriginal infants, Western Australia



1980-92 –before any health promotion or fortification

1993-95 –health promotion of folic acid supplement use

1996-06 –health promotion & voluntary fortification

2007-09 –transition period

2010-14 –post-mandatory fortification

Fetal Alcohol Spectrum Disorder



What is FASD?

- FASD is a condition characterised by
 - severe, pervasive, neurodevelopmental impairment
 - due to prenatal exposure to alcohol
- The diagnosis of FASD has two sub-categories:
 - FASD with 3 sentinel facial features
 - FASD with < 3 sentinel facial features

Australian Guidelines for Diagnosis of FASD:

<http://alcoholpregnancy.telethonkids.org.au/australian-fasd-diagnostic-instrument/australian-guide-to-the-diagnosis-of-fasd/>

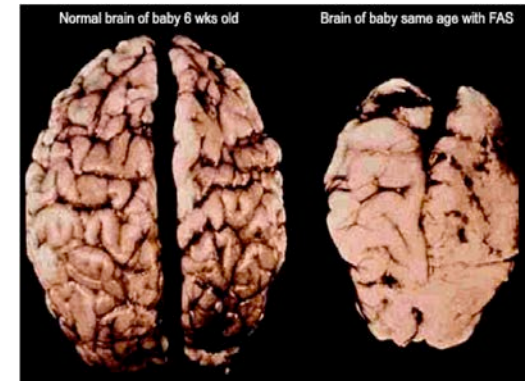
FASD with 3 sentinel facial features



Prenatal alcohol exposure



Three sentinel facial features



Neurodevelopmental impairment in 3+ domains

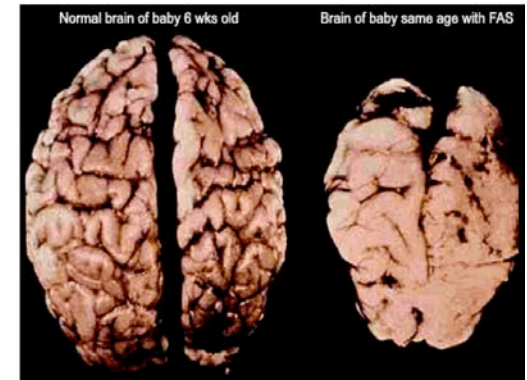
FASD with < 3 sentinel facial features



Prenatal alcohol exposure



0, 1 or 2 sentinel facial features



Neurodevelopmental impairment in 3+ domains

Sentinel facial features



- Short palpebral fissure
- Smooth philtrum
- Thin upper lip



FASD neurodevelopmental impairment

Severe impairment in 3 or more of these domains:

- Brain structure/neurology
- Motor skills
- Cognition
- Language
- Academic achievement
- Memory
- Attention
- Executive function, impulse control and hyperactivity
- Affect regulation
- Adaptive behaviour, social skills or communication





What disabilities can result from FASD?

- Behaviour problems
- Speech and language difficulties
- Learning difficulties
- Cognitive problems
- Executive functioning problems
- Motor and sensory difficulties
- ADHD; Autistic features

Secondary problems – school, mental health, drug and alcohol problems, justice



Evidence

- **Alcohol use in pregnancy**
 - 7-20% women drink at high risk levels
 - 57% women drink some alcohol in pregnancy
- **Health professionals' surveys:**
 - majority don't ask about alcohol use in pregnancy;
 - most acknowledge no alcohol safest option;
 - majority want professional development;
 - < 20% knew diagnostic criteria for FAS



FASD birth prevalence

Australian Paediatric Surveillance Unit, 2001-2004

- 92 cases FAS; 0.06/1000; majority Aboriginal
- Increase in notifications over study period

WA Register of Developmental Anomalies, 1980-2010

- 210 cases; 0.26/1000; majority Aboriginal
- **Two-fold increase** in FASD in 2000-2010 in both Aboriginal and non-Aboriginal children

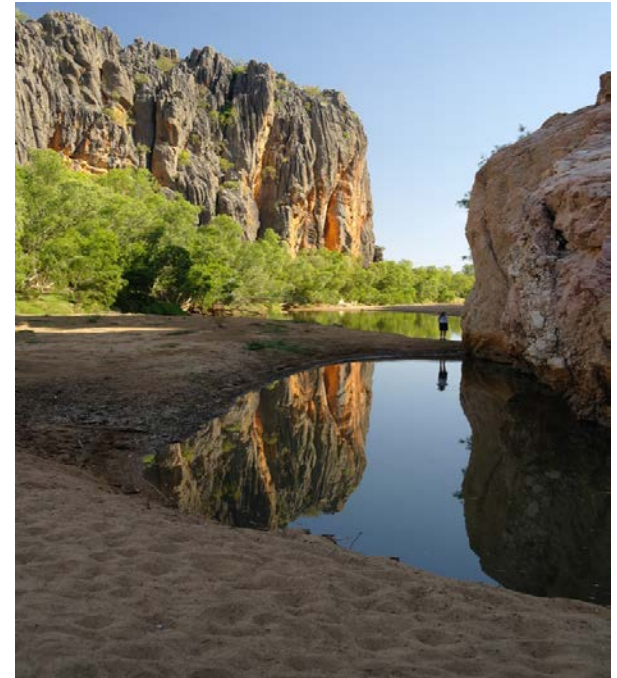
Increase due to improved diagnosis and notification
Still under-recognised

Elliott et al. Arch Dis Child 2007

Mutch et al. J Paediatr Child Health 2014

Lililwan project

- Aboriginal initiated and led
- All 7-8yos in Fitzroy Valley
- 108/134 (81%) assessed
- 55% alcohol use in pregnancy
- FASD in 19%



Fitzpatrick et al. J Paediatr Child Health 2015; unpublished



Primary prevention

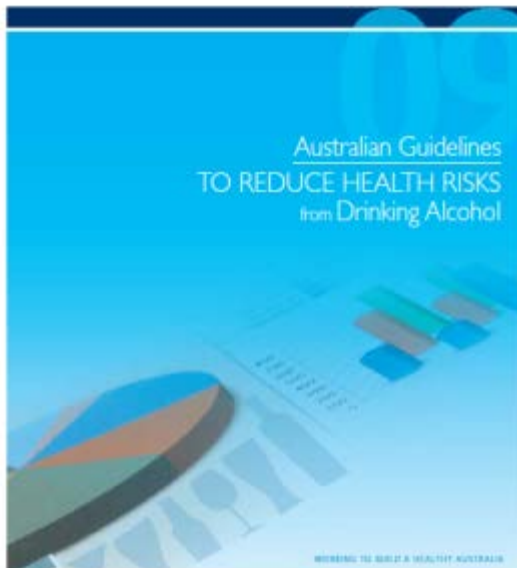
- Evidence
- Engagement
- Evaluation



The problems..

- 50% women drink alcohol in pregnancy
- Role of alcohol in society
- Concerns about stigmatisation
- Discomfort asking about alcohol
- Conflicting messages
- Unplanned pregnancies – 40-50%
- Continual education

Australian Guidelines, 2009



Maternal alcohol consumption can harm the developing fetus or breastfeeding baby.

A. For women who are pregnant or planning a pregnancy, **not drinking is the safest option.**

B. For women who are breastfeeding, not drinking is the safest option.

<http://www.nhmrc.gov.au/files/nhmrc/file/publications/synopses/ds10-alcohol.pdf>



Primary prevention

Education and support for health professionals

- Ask, assess and advise at every opportunity
- Brief intervention and support
- Assist with referral for women with problems
- Early diagnosis of FASD – important for next pregnancies

Health promotion

- general population
- high risk



Alcohol and Pregnancy

Evaluation of resources

- are used by health professionals
- are clinically relevant
- have had an impact on health professionals' practice
- 48% have changed or intended to change their practice



Prevention



- ***Marulu FASD Prevention Strategy*** – the Fitzroy Valley communities have bold goal to “Make FASD History” by reducing alcohol use in pregnancy
- 55% women drinking in pregnancy in 2003
- ~35% women drinking in pregnancy in 2013
- **Target <10%** by 2018
- Expansion to Pilbara



Conclusions

- Early childhood development starts before birth
- Population approach to a good start in life
 - Evidence
 - Engagement
 - Evaluation
- Primary prevention of birth defects is possible
- Recognition and management of effects of birth defects





Visit our website:

<http://alcoholpregnancy.telethonkids.org.au/>





WA Register of Developmental Anomalies

- Established in 1980
- Statutory collection
- Active ascertainment
- High level of completeness and accuracy
- Active engagement with:
 - Consumers and community
 - Clinicians
 - Government
 - Researchers

- 33,000 births a year in Western Australia
- 6% birth defects



http://kemh.health.wa.gov.au/services/register_developmental_anomalies/