Population approaches to the primary prevention of birth defects

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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

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-conceptional 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

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

Sales of folic acid supplements

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

Bower et al, PPE 2004;18:277-281; Data from the WARDA
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

Increase due to folate in fortified bread

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

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
- **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:
FASD with 3 sentinel facial features

Prenatal alcohol exposure + Three sentinel facial features + Neurodevelopmental impairment in 3+ domains

Normal brain of baby 6 wks old

Brain of baby same age with FAS
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
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.

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
• **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:
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