Can we improve the prevention and detection of congenital abnormalities? An audit of early pregnancy care in New Zealand

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Abstract

Aim To determine whether there were “quality gaps” in the provision of care during pregnancies that resulted in a perinatal death due to congenital abnormality.

Method Perinatal deaths from congenital cardiovascular, central nervous system or chromosomal abnormality in 2010 were identified retrospectively. Data were extracted by retrospective clinical note review and obtained by independent review of ultrasound scans.

Results There were 137 perinatal deaths due to a congenital cardiovascular (35), central nervous system (29) or chromosomal abnormality (73). First contact with a health professional during pregnancy was predominantly with a general practitioner. First contact occurred within 14 weeks in 85% of pregnancies and there was often a significant delay before booking. Folate supplements were taken by 7% pre-conceptually and 54% of women in the antenatal period. There were 20 perinatal deaths from neural tube defects that could potentially have been prevented through the use of pre-conceptual folate. Antenatal screening was offered to 75% of the women who presented prior to 20 weeks and 84% of these undertook at least one of the available antenatal screening tests. Review of ultrasound images found five abnormalities could have been detected earlier.

Conclusion Delay in booking or failure to offer screening early were the most common reasons for delay in diagnosis of screen detectable abnormalities. The preventative value and timing of (pre-conceptual) folate needs emphasis.

There were 704 perinatal deaths in New Zealand in 2010, and 30% of these perinatal deaths were due to congenital abnormalities.¹ Congenital abnormality is the most common cause of perinatal death in New Zealand, and therefore a review of the care received during pregnancy was designed to identify areas for improvement.

It was hypothesised that a number of congenital abnormalities could be prevented if folate was taken prenatally. It was also hypothesised that a number of chromosomal abnormalities and neural tube defects could be detected earlier through current regimens of antenatal screening in the first or second trimester.

Antenatal screening has been an accepted part of antenatal care in New Zealand since 1968. In February 2010 the National Screening Unit introduced a new guideline for routine antenatal screening, to be offered to all pregnant women, which includes a nuchal translucency scan and blood test (levels of plasma protein-A and beta human chorionic gonadotrophin) between 11 and 13 weeks gestation.
If women are unable to access the first trimester screening, a second trimester blood test (levels of beta human chorionic gonadotrophin, alpha fetoprotein, unconjugated oestriol and inhibin A) between 14 and 20 weeks is also available.

The first and second trimester screening combination of tests is calibrated to identify an increased risk of Trisomy 21, with a lower sensitivity to identify an increased risk of other trisomies and chromosomal abnormalities (trisomy13,18, triploidy, Turner’s and Klinefelter’s syndromes).

Second trimester serum screening will also identify an increased risk of open neural tube defects (anencephaly, acrania, spina bifida and encephalocele). All women are offered a fetal anatomy scan at around 20 weeks gestation to check for fetal abnormalities.

Antenatal screening and ultrasound are key tools for identifying congenital abnormalities early in pregnancy. Early identification gives parents a greater number of options for treatment, and may reduce the number of late terminations of pregnancy (after 20 weeks gestation).

Late terminations are associated with increased risks to the mother, greater maternal distress and additional requirements for statutory registration of death.2

Method

Perinatal deaths resulting from cardiovascular system, central nervous system or chromosomal congenital abnormality during 2010 were identified from the Perinatal and Maternal Mortality Review Committee (PMMRC) dataset.

The PMMRC dataset of perinatal deaths is a compilation of data submitted by Lead Maternity Carers (LMCs), clinicians, PMMRC District Health Board (DHB) local coordinators, death notifications and some additional data from births deaths and marriages (BDM).

The perinatal deaths included in the audit occurred between 1 January and 31 December 2010. For fetal deaths, the date of birth is used as ‘date of death’. Only fetuses and babies who died from 20 weeks gestation up to 27 days after birth are included in this audit. This means that a significant number of fetuses are not included in this audit as the pregnancy would have ended prior to 20 weeks and were therefore not within the scope of the PMMRC.

The classification system of cause of death that has been adopted by the PMMRC is the Perinatal Society of Australia and New Zealand (PSANZ) system of classification (PDC).3

Perinatal deaths included in this audit were classified as central nervous system (PDC 1.1), cardiovascular (PDC 1.2) or chromosomal abnormalities (PDC 1.5). These sub-classifications of congenital abnormalities are the most likely to be detected by first and second trimester antenatal screening. The hospital notes, Lead Maternity Carer (LMC) notes and General Practitioner (GP) notes were requested where applicable for each of the pregnancies included in the audit.

An audit tool was developed to gather key demographic data and information on pregnancy care. The notes for each of the women were reviewed and the data points for each woman entered into an Excel spread sheet.

A review of ultrasound images was undertaken. Using the ultrasound reports included in the women’s notes we were able to identify women who had scans between 10 weeks and the gestation at which the abnormality was detected.

Ten weeks was used as the cut off as it is the lower limit at which most congenital abnormalities can be identified on ultrasound. Women who had a scan after 10 weeks which was reported as normal were included in the ultrasound audit [n=82/137 (60%)].

Static images for the ultrasounds were reviewed by one specialist with expertise in ultrasound and maternal fetal medicine using the Picture Archiving and Communication System (PACS) or in a DVD format. The reviewer was aware that the pregnancy had ended in a perinatal death due to congenital
abnormality; however the type of abnormality was not known. None of the ultrasound scans that were reviewed had been reported by the reviewer.

Results

137 of the 211 perinatal deaths from congenital abnormality reported to the PMMRC were identified and confirmed as cardiovascular system (26%), central nervous system (21%) or chromosomal abnormalities (53%).

Of the 137 women included in the audit, six sets of LMC clinical notes were not available as they had either been given to the woman or the midwife had left the country. In three cases there was no specific reason for the midwife not to have retained a copy of the notes.

Table 1 shows the maternal demographic data by perinatal death classification. Median BMI was higher for women with a baby with a central nervous system abnormality compared to women with other congenital abnormalities but this difference was not statistically significant (p=0.13).

Table 1. Maternal demographic data

<table>
<thead>
<tr>
<th>Perinatal death classification (PSANZ PDC)</th>
<th>Total</th>
<th>n=35</th>
<th>n=29</th>
<th>n=73</th>
<th>n=137</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prioritised ethnicity</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Maori</td>
<td>8</td>
<td>22.9</td>
<td>5</td>
<td>17.2</td>
<td>9</td>
</tr>
<tr>
<td>Pacific Peoples</td>
<td>9</td>
<td>25.7</td>
<td>1</td>
<td>3.4</td>
<td>13</td>
</tr>
<tr>
<td>Indian</td>
<td>0</td>
<td>0.0</td>
<td>0</td>
<td>0.0</td>
<td>2</td>
</tr>
<tr>
<td>Other Asian</td>
<td>1</td>
<td>2.9</td>
<td>7</td>
<td>24.1</td>
<td>8</td>
</tr>
<tr>
<td>Other</td>
<td>1</td>
<td>2.9</td>
<td>5</td>
<td>17.2</td>
<td>6</td>
</tr>
<tr>
<td>NZ European</td>
<td>16</td>
<td>45.7</td>
<td>11</td>
<td>37.9</td>
<td>35</td>
</tr>
<tr>
<td>Mother’s age</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>&lt;20</td>
<td>4</td>
<td>11.4</td>
<td>2</td>
<td>6.9</td>
<td>2</td>
</tr>
<tr>
<td>20-24</td>
<td>12</td>
<td>34.3</td>
<td>10</td>
<td>34.5</td>
<td>9</td>
</tr>
<tr>
<td>25-29</td>
<td>8</td>
<td>22.9</td>
<td>5</td>
<td>17.2</td>
<td>10</td>
</tr>
<tr>
<td>30-34</td>
<td>5</td>
<td>14.3</td>
<td>9</td>
<td>31.0</td>
<td>17</td>
</tr>
<tr>
<td>35-39</td>
<td>5</td>
<td>14.3</td>
<td>3</td>
<td>10.3</td>
<td>21</td>
</tr>
<tr>
<td>40+</td>
<td>1</td>
<td>2.9</td>
<td>0</td>
<td>0.0</td>
<td>14</td>
</tr>
<tr>
<td>BMI (median (IQR))</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>29.4 (21.7-34.6)</td>
<td>23.1 (20.5-28.7)</td>
<td>24.3 (21.9-27.4)</td>
<td>24.9 (21.6-31.2)</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Folate supplements were reported to have been taken by only 7% of women pre-conceptually and 54% of women antenatally. Of the 21 whose babies were diagnosed with neural tube defects (congenital abnormalities amenable to folate use) only one out of 21 was recorded as taking pre-conceptual folate and 13 out of 21 (62%) antenatal folate.

A majority of women [93 (68%)] were first seen by their GP during pregnancy, while 30 women (22%) had their first contact with a self-employed LMC, eight were first
seen in hospital or by a school nurse and first contact was unknown for seven of the women.

Figure 1 shows the association between gestation in weeks at which the mother was first seen by a health professional on the x axis compared to the gestation in weeks when she booked with a LMC. Dashed lines are shown at 10 weeks when booking is advised and solid lines at 14 weeks which is the final gestation for first trimester screening.

Overall 114 women (83%) were seen by a health care provider before 14 weeks, while only 90 women (66%) booked with a LMC before 14 weeks.

Figure 1. Scatter plot of gestation at first health professional visit and gestation at booking with LMC among women whose babies died of PDC 1.1, PDC 1.2 and PDC 1.5 in New Zealand 2010

Figure 2 shows an overview of the gestation at first contact and screening history of the study population. First and second trimester screening are reported together as only 17 women had second trimester serum screening and some of these women also had a nuchal translucency in first trimester.

Of the 82 who took up the offer of first/second trimester screening 27 had a nuchal translucency scan alone, 38 had combined first trimester screening, 13 had a nuchal translucency scan and second trimester bloods, and four had second trimester bloods alone.
Figure 2. Outcomes of first and second trimester nuchal translucency and serum screening among perinatal related deaths from central nervous system (PDC 1.1), cardiovascular system (PDC 1.2) and chromosomal (PDC 1.5) congenital abnormalities in New Zealand 2010

There were eight women who had a nuchal translucency scan but did not have a first trimester blood test done so no risk was reported (2010 guidelines prevent ultrasound providers from reporting the risk based on the nuchal translucency measurement alone).

Of these eight women, two had second trimester blood tests; low risk in one case and an increased risk result in the other case. The increased risk pregnancy was found to be Trisomy 21 and the only abnormality in this group of eight that was amenable to screening.

Table 2 shows a breakdown of the screening results by abnormality and gives false negative rates. A false negative was defined when a woman was deemed by appropriate screening to be low risk for the identified abnormality.

Trisomy 21 had a low false negative rate in this sample with only one out of the nine Trisomy 21 pregnancies who underwent screening receiving a low risk result. While the false negative rate for all potentially screen detectable abnormalities is shown it should be noted that the only true false negatives are those for Trisomy 21 which is the abnormality the screening test is currently calibrated for.

* Includes 6 cases of spina bifida who did NOT have second trimester serum testing so would not have been detected.
Table 2. First and second trimester screening results by congenital abnormality group

<table>
<thead>
<tr>
<th>Type of abnormality</th>
<th>Total</th>
<th>First or second trimester screening</th>
<th>Increased risk result</th>
<th>False negative screen*</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>n</td>
<td>n</td>
<td>%</td>
<td>n</td>
</tr>
<tr>
<td>CNS abnormalities</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Anencephaly/acrania**</td>
<td>10</td>
<td>4</td>
<td>40</td>
<td>3</td>
</tr>
<tr>
<td>Spina bifida</td>
<td>9</td>
<td>8†</td>
<td>89</td>
<td>0</td>
</tr>
<tr>
<td>Encephalocele</td>
<td>2</td>
<td>0</td>
<td>0</td>
<td>-</td>
</tr>
<tr>
<td>Other CNS*</td>
<td>14</td>
<td>7</td>
<td>50</td>
<td>-</td>
</tr>
<tr>
<td>Cardiac abnormalities</td>
<td>All cardiac*</td>
<td>29</td>
<td>18</td>
<td>62</td>
</tr>
<tr>
<td>Chromosomal abnormalities</td>
<td>Trisomy 21**</td>
<td>18</td>
<td>9</td>
<td>50</td>
</tr>
<tr>
<td></td>
<td>Trisomy 20**</td>
<td>1</td>
<td>1</td>
<td>100</td>
</tr>
<tr>
<td></td>
<td>Trisomy 18**</td>
<td>19</td>
<td>12</td>
<td>63</td>
</tr>
<tr>
<td></td>
<td>Trisomy 13 (includes 13/18)**</td>
<td>6</td>
<td>4</td>
<td>67</td>
</tr>
<tr>
<td></td>
<td>Turner syndrome**</td>
<td>3</td>
<td>2</td>
<td>67</td>
</tr>
<tr>
<td></td>
<td>Klenefelter's syndrome**</td>
<td>2</td>
<td>1</td>
<td>50</td>
</tr>
<tr>
<td></td>
<td>Other chromosomal*</td>
<td>18</td>
<td>12</td>
<td>67</td>
</tr>
</tbody>
</table>

*Other CNS, all cardiac or other chromosomal are not screen detectable. However in some cases the first trimester screening will show an increased result due to a large nuchal translucency measurement indicating illness in the fetus.

** First trimester nuchal translucency with or without first or second trimester serum screening  
† Only two of eight cases of spina bifida had second trimester serum screening (Spina bifida is only detectable by second trimester serum screening or ultrasound in the second trimester and ultrasound is the primary method for detecting spina bifida).

The final diagnosis was made significantly earlier in screen detectable abnormalities where antenatal screening occurred in either the first or second trimester [median gestation 19.5 weeks (IQR 19-21)] compared to when no screening was done [21 weeks (IQR 19-25)], p=0.02.

Termination was offered earlier in pregnancy when the pregnancy resulted in a screen detectable congenital abnormality and the mother had undergone screening in the first or second trimester [median gestation of 20 weeks (IQR 19-21) compared to 21 weeks (IQR 20-24) among unscreened women], although this difference was not statistically significant.

126 women (92%) had an anatomy scan. The median gestation at the anatomy scan was 19 weeks, ranging from 19 to 26 weeks. Of the 126 women who had an anatomy scan 96 (76%) were reported as abnormal, 16 (13%) as unclear and 14 (11%) were reported as low risk. The unclear group was made up predominantly of scans where
part of the fetal anatomy was not adequately seen which could have been due to imaging issues or to abnormality.

Eighty-two cases where a valid scan was available (131 ultrasound scans) were reviewed. Twenty five scans met the criteria for review but were unable to be obtained. The review of available scans identified five cases where the reviewer was able to identify the abnormality earlier. This included one trisomy 18, one triploidy, one central nervous system abnormality and two cardiovascular system abnormalities. There was an additional case where the images were not available where it was thought that there was potential for the abnormality to have been detected earlier due the severe nature of the abnormality.

There were four sets of anatomy scan images that were not available for review. Of the 20 sets of anatomy scan images that were available, seven were missing key views which meant that the scan should not have been reported as complete.

Discussion

There were 137 perinatal deaths where the primary antecedent cause of death was cardiovascular, central nervous system or chromosomal congenital abnormality in 2010. These constitute a significant portion (19%) of the 704 perinatal deaths during 2010 in New Zealand.

The purpose of this audit was to identify areas where there is potential to improve care for women with babies with congenital abnormalities and to reduce the number of perinatal deaths due to these types of congenital abnormalities firstly through prevention and secondly through earlier detection.

Only one of the 21 mothers who had a pregnancy that resulted in a neural tube defect was recorded as having taken folic acid pre-conceptually and 13 were recorded as having taken it antenatally.

The Ministry of Health recommends that all women who are planning a pregnancy or who are pregnant take 0.8mg of folic acid (5mg if in a higher risk group). If taken for at least one month prior to conception and during the first three months of pregnancy, folate can reduce the incidence of neural tube defects (risk ratio for folate use for reducing NTDs 0.28, 95% confidence interval 0.15 to 0.52).

Pre-pregnancy counselling including optimising treatment of medical conditions, identifying personal and family history of congenital defects, advising on folate prophylaxis, smoking cessation and educating about the potential value of booking early with an LMC are all important ways to identify at risk pregnancies and to reduce risk of congenital abnormalities.

A delay between first contact with a health care professional and booking with a lead maternity carer (LMC) was identified. In a majority of cases the first contact during the pregnancy was with a general practitioner (or general practice nurse). This first contact occurred before the cut off for first trimester screening (13 weeks and six days) in 83% of pregnancies examined, however only 66% of women went on to book with an LMC before 14 weeks.

This highlights a need to facilitate booking with a LMC or for GPs to take the responsibility for providing antenatal screening. If GPs continue to provide first line
antenatal care, they should be targeted for first trimester antenatal screening education.

In February 2010 the antenatal screening guidelines in New Zealand changed. Prior to 2010 the main antenatal screening tests for congenital abnormalities were the nuchal translucency and anatomy scans. In February 2010 new guidelines were implemented that required all eligible pregnant women to be offered a nuchal translucency scan combined with a blood test prior to 14 weeks gestation.

Women who presented after 14 weeks could be offered the 2\textsuperscript{nd} trimester blood test up to 20 weeks gestation. There was a range of screening test combinations undertaken by the women included in this audit, which may reflect the changes that were made to the screening programme during this period, although it may also reflect women’s choices. It would be useful to evaluate whether there continues to be variation in antenatal screening options performed.

First and second trimester antenatal screening is calibrated for maximal sensitivity for Trisomy 21, although it does identify risk for other chromosomal abnormalities. The calibration of the screening optimising diagnosis of Trisomy 21 is evidenced by the higher false negative rates in pregnancies which resulted in Trisomy 18 or Trisomy 13 in this review.

Screening could be calibrated for greater detection of Trisomy 18 and Trisomy 13, but the costs and benefits of these additional investigations would need to be evaluated. The true false negative rates for any chromosomal abnormality can only be assessed by a review of all cases of congenital abnormality. To do this would require an improved register of congenital abnormalities in New Zealand.

The efficacy of the anatomy scan as a tool for identifying abnormalities is partly due to high uptake of this test (92% in this review), and partly due to a high rate of detection (76% of the anatomy scans in this data set were reported as abnormal).

To be most effective, the anatomy scan should be done at 20 weeks which is a recent change from 18 weeks and reflects in part the changing maternal habitus as increasing obesity has made it more difficult to obtain the required images.

Having the anatomy scan later increases the chance of an abnormality being detected in all women, although this needs to be weighed against the effects of a later diagnosis.

The anatomy scans that on review were determined to be incomplete but were reported as completed are an area of concern. With usual practice a sonographer scans the patient and captures the required images and then a radiologist reviews and reports on the static images. When the required images are not captured the anatomy of the baby is not able to be adequately assessed.

Ensuring that all required images are obtained and getting women to return for a further scan should the required images not be obtained are important steps to improving detection of abnormalities. Failure to obtain a standard view may be an indicator that there is an abnormality.
There were five cases where the ultrasound reviewer was able to detect the abnormality earlier. These cases suggest ultrasound is an area where the detection of congenital abnormalities could be improved.

The failure to retain static images by ultrasound operators was identified as an issue. Given continued improvements in electronic storage, the reduction in cost of this service and importance to audit and review, ultrasound providers should be retaining copies of ultrasound scans. Further, this practice aligns with retention of clinical notes by clinicians in other specialties.

Of the 137 women included in the audit there were six sets of midwifery notes that were not available for review. All lead maternity carers are legally required to retain a copy of pregnancy notes for 10 years.6

**Conclusion**

This audit aimed to determine whether there were quality gaps in the care of women whose babies died from congenital abnormality. This included the hypotheses that a number of congenital abnormalities could be prevented if folate was taken prenatally and that a number of chromosomal abnormalities and neural tube defects could have been detected earlier.

Preconceptional folate supplements were not widely reported as used across all types of abnormalities, levels of folate supplement use reported increased antenatally. It is likely that given the low levels of preconceptional folate use reported that a number of the neural tube defects could have been prevented had folate supplements been taken preconceptually.

A number of the abnormalities could potentially have been detected earlier this includes 23% of women who saw a health care practitioner before the cut off for either first or second trimester screening and who were not offered screening.

There were also three neural tube defects that could potentially have been detected earlier including an anencephaly that was not detected at the nuchal translucency scan and two spina bifidas that were not detected during the second trimester screening. This may reflect the fact that second trimester screening is no longer optimised for detecting spina bifida as not all spina bifidas are screen detectable.

Similarly there were ten chromosomal abnormalities that could potentially be detected by screening but the screening algorithms are not optimised to detect them. There was one case of Trisomy 21 that was a false negative.

**Recommendations where improvements could be made based on the findings include:**

- All women should receive pre-conceptual counselling to optimise maternal health, identify obstetric or familial risk factors, discuss current medical conditions and refer to specialists as required.

- A media campaign for pre-conceptual folate is required along with further investigation of the fortification of bread with folate.
• Education of all women is required about the importance of booking with a LMC before 10 weeks.

• Education and support should be offered so that primary care providers are able to effectively offer first trimester screening, interpret screening results and facilitate expeditious booking with an LMC.

• If screening has not already been arranged then LMCs should offer all women first or second trimester screening, as required by the Ministry of Health.

• There should be a review of the current algorithms used in New Zealand’s first and second trimester screening programme and consideration of the cost benefit of using algorithms calibrated for maximal sensitivity for all chromosomal abnormalities.

• There should be a review of the efficiency and adequacy of the antenatal screening program’s guidelines for reporting results for nuchal translucency in a patient who has not had a serum sample taken to avoid delays in reporting risk from the nuchal scan.

• False negative screening tests should be reviewed by the screening unit.

• All LMCs should document pre-conceptual folate and antenatal folate use including when the woman commenced taking folate and the dose.

• Ultrasound services should retain copies of all ultrasounds and audit their images to ensure accurate measurements are obtained during scanning, in particular during the nuchal translucency scan.

• Enhancement of the current birth defects register to include congenital abnormalities where a perinatal death occurred.

Competing interests: None identified.

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References:


The rate of all major congenital abnormalities other than neural tube defects and genetic syndromes diagnosed up to the eighth month of life was 14.7/1000 informative pregnancies in the vitamin group and 28.3/1000 in the trace element group; relative risk 1.95 (1.23 to 3.09); difference, 13.6/1000. The rate of some congenital abnormalities was lower in the vitamin group than in the trace element group but the differences for each group of abnormalities were not significant. You are going to email the following Prevention of congenital abnormalities by periconceptional multivitamin supplementation. Your Personal Message. Topics.