BIRTH DEFECTS – THE STATE OF AWARENESS AMONGST MOTHERS

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ABSTRACT

Birth defects have in recent years become the major cause of perinatal morbidity and mortality. The incidence of birth defects is between 2% to 6% of all live births. However, from a survey carried out in Singapore General Hospital (SGH), the majority of mothers enter pregnancy without realising the risks that they or their offspring may face. Only 20% to 25% of mothers were aware of the actual incidence of birth defects, and only 2.5% – 10% knew that the risk of Down Syndrome (DS) increases with maternal age. However, the use of ultrasound is now well accepted and is considered a necessary investigation by 90% of the mothers surveyed.

Keywords: *mothers, awareness, birth defects, Down syndrome, ultrasound.*

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INTRODUCTION

In Singapore, where the age of childbearing has definitely increased while the size of the family unit has decreased, every pregnancy is now expected to progress without any problems, resulting in a perfect baby. In reality, the incidence of congenital birth defects is still in the region of 2% to 6%[1] and is the largest contributor to perinatal and neonatal mortality. Mothers of today cannot choose to ignore the issue of birth defects and steps are necessary to increase their knowledge and awareness.

This survey was carried out within SGH to determine how knowledgeable our pregnant mothers were with regards to the problem of chromosomal and birth defects in pregnancy as it was felt subjectively during our counselling sessions that their knowledge was either inadequate or inaccurate.

METHODOLOGY

A questionnaire was designed to survey two different groups of pregnant mothers within SGH on their awareness of a woman's risk of having a child with either Down Syndrome (DS) or structural birth defects and their knowledge of the currently available prenatal diagnostic procedures. Their perception of what constituted adequate antenatal care and the role of ultrasound in obstetric management was also tested. This survey was carried out between 1st January and 30th June 1992.

One hundred and fifty postnatal mothers who had delivered healthy babies were given the questionnaire within the first few days of their puerperium prior to their discharge. Sixty antenatal mothers who had been counselled for prenatal diagnosis and were about to undergo an invasive procedure were given a similar questionnaire as a comparative group in this study.

The questionnaire used was the multiple choice form used in the first awareness survey[2].

Mode of counselling for the antenatal group

In the antenatal group, the mothers usually accompanied by their spouses, would have had a 30-minute private counselling session with a trained counsellor. A video tape presentation providing basic information about genetic and chromosomal disorders eg Thalassaemia and Down Syndrome is viewed by the couple to give them an understanding as to the need for prenatal diagnosis. The maternal age specific risk for DS and other chromosomal aberrations is explained to the couple.

The two modes of invasive prenatal diagnostic procedures, chorionic villous sampling and amniocentesis, are then discussed with the couple. The advantages and disadvantages of both procedures are explained to the couple.

Consent is then obtained from the couple after they have understood the counselling advice given and have agreed to have a prenatal diagnostic procedure performed.

The antenatal (AN) mothers were surveyed on the day of their scheduled procedure. Postnatal (PN) mothers on the other hand have no formal counselling and were surveyed either on the day of their delivery or one day later.

SURVEY FINDINGS AND POSTULATIONS

General Risk

The survey revealed that the majority of mothers held the erroneous belief that the general risk of having an abnormal baby was less than 1 in 500. Only 26% of postnatal mothers and 20% of antenatal mothers thought the risk was 1 in 100 or more which was the closest to the actual incidence of 2% to 6% foetal anomaly in total number of babies delivered (Table I).

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Table I – Perceived general risk of having an abnormal baby.

Risk	PN Mothers	AN Mothers
1 in 100 or more	26.0%	20.0%
1 in 500	16.6%	23.3%
1 in 1000	28.6%	30.0%
1 in 10,000	21.3%	10.0%

Down Syndrome

While only 15.3% of postnatal mothers knew that Down Syndrome was a chromosomal disorder of Trisomy 21, another 68% of this group knew that DS was either a genetic disorder or resulted in a mentally retarded child.

Antenatal mothers had already been extensively counselled as to what DS was, and therefore were not asked this question.

Table II – Perceived risk of Down Syndrome(DS) with changes in the maternal age

Risk of DS with maternal age	PN Mothers	AN Mothers
No change	11.3%	18.3%
Increased	80.7%	71.7%
Decreased	4.0%	10.0%

80.7% and 71.7% of postnatal and antenatal mothers respectively were aware that the risk of DS increases with the maternal age. It was disappointing to note that the antenatal mothers who were specifically counselled with regards to maternal age related risk of chromosomal problems did not achieve 100% (Table II).

Chromosomal Abnormalities

The risk of chromosomally abnormal children does not depend on the number of children a woman has, but on the maternal age and the presence of other high risk factors in the family history.

This was correctly perceived by 66.0% and 76.7% of postnatal and antenatal mothers respectively – which was not unexpected as most mothers thought that once they had a normal child they would be at low risk of having an abnormal baby (Table III).

Table III – Perceived risk of chromosomal abnormalities with increasing number of children

Risk with Increasing parity	PN Mothers	AN Mothers
No change	66.0%	76.7%
Increased	20.7%	5.0%
Decreased	6.6%	10.2%

Structural Abnormalities

As the risk of chromosomal abnormalities increases with maternal age, the incidence of their associated structural abnormalities will also increase. Although the specific incidence of structural abnormalities which occur de novo does not change with age, the overall incidence of structural abnormalities will still increase with maternal age.

Approximately half of both groups of mothers thought correctly that the risk of structurally abnormal children increased with maternal age (Table IV).

Table IV – Perceived risk of having babies with structural abnormalities changes with maternal age

Risk with increasing maternal age	PN Mothers	AN Mothers
No change	32.7%	53.3%
Increased	59.3%	41.7%
Decreased	4.7%	3.3%

Approximately 60% of both groups of mothers thought that the risk of having an abnormal child did not change with increasing parity probably again due to the fact that they already have previous normal pregnancies (Table V).

Table V – Perceived risk of having babies with structural abnormalities with increasing parity.

Risk with increasing parity	PN Mothers	AN Mothers
No change	68.6%	63.3%
Increased	16.7%	15.0%
Decreased	4.0%	5.0%
No response	10.7%	16.7%

Use of ultrasound in pregnancy

Table VI – Perceived necessity of an ultrasound scan for every pregnant woman

Is ultrasound a necessity?	PN Mothers	AN Mothers
Yes	83.3%	95.0%
No	13.3%	1.6%

Perceived detection rate of foetal abnormalities on ultrasound

Realistically, the detection of foetal abnormalities depends on many variables. However under ideal circumstances one can realistically expect to detect 75% of all foetal abnormalities[3-5]. Obviously the mothers sampled were not sure of the answer as there was a fairly even distribution. 25.3% postnatal and 30% antenatal mothers thought the ultrasound scan could only detect less than 50% of the major abnormalities while at the other extreme 19.5% and 18.3% expected the ultrasound scan to detect 90 to 100% of all abnormalities respectively (Table VII).

Table VII – Perceived rate of detection of major abnormalities by ultrasound.

Rate of detection	50%	60%	70%	80%	90%	100%
PN Mothers	25.3%	15.3%	16.0%	20.8%	12.7%	6.8%
AN Mothers	30.0%	8.3%	10.0%	20.0%	13.3%	5.0%

Timing of ultrasound scans in pregnancy

Forty-eight percent and 61.7% of postnatal and antenatal mothers thought that the best time to scan was at around 12 weeks of gestation, while only 18.0% and 11.7% of the respective mothers felt that the best time to scan was at 20 weeks of gestation[6,7] (Table VIII).

This is a rather surprising finding as all mothers booked in the department have a routine antenatal screening ultrasound scan performed for them at around 20 weeks if they have been booked in the first trimester. In the antenatal mothers group, this erroneous idea may have arisen as a first trimester dating ultrasound scan was always done prior to counselling and the subsequent prenatal diagnostic procedure[8].

Table VIII – Perceived optimal time for structural screening ultrasound scan for foetus.

Gestation	PN Mothers	AN Mothers
12 weeks +	48.0%	61.7%
16 weeks +	25.0%	8.3%
20 weeks +	18.0%	11.7%
24 weeks +	6.0%	1.7%
> 28 weeks	2.0%	1.7%
Not necessary	0%	1.7%

Gestation at which first visit was thought best

It was heartening to note that 70.0% of postnatal mothers and 83.3% of antenatal mothers thought that the first visit to a doctor should occur two weeks after the missing the expected menses. Only 25.3% of postnatal and 13.3% of antenatal mothers felt that the first consultation could wait till some time between the third and fourth month of gestation (Table IX).

This perhaps again is a reflection of the types of mothers in the two groups. The postnatal mothers group were those who mostly had uneventful antenatal follow up while the antenatal mothers were usually older, more educated and were more likely to have had a history of subfertility, and thus were understandably keen to have the best possible care from very early on in their pregnancy.

Table IX – Perceived best time for the first consultation in pregnancy.

Gestation	6/52	12-20/52	20-28/52	28-36/52	>36/52
PN Mothers	70.0%	25.3%	2.0%	0.0%	0.6%
AN Mothers	83.3%	13.3%	0%	1.7%	1.7%

Knowledge of prenatal diagnostic services

71.3% of postnatal mothers knew that other services aside from ultrasound were available to help determine the normality of their foetus prior to delivery. 72.6% were aware of amniocentesis but the majority had never heard of the terms chorionic villous sampling, cordocentesis or karyotyping.

Attitudes towards foetal abnormalities

The vast majority, 95% antenatal and 91.3% postnatal mothers, would prefer to know if they were bearing an abnormal child. 78.3% and 77.3% of these mothers respectively would choose to terminate their pregnancy if they knew that their baby had a fatal or complex abnormality.

DISCUSSION

The birth of an abnormal baby would inevitably come as a shock to its parents. The public at present has much better access to the latest knowledge in antenatal care from newspapers, public forums, mothercraft features on the radio and television as well as the numerous books on pregnancy care, and lastly their family practitioner or obstetrician.

In recent years, foetal abnormality has emerged as the major cause of perinatal mortality as well as a significant contributor to childhood morbidity. Many studies have shown that the risk of having an abnormal baby at birth is in the range of 2% to 6% of total births[1]. Every couple who plans for their family can no longer ignore the possibility of their offspring being the unfortunate few.

Pregnant mothers were surveyed because it was felt that they would be the group with the highest likelihood of having the most current and accurate knowledge on the state of prenatal diagnosis on birth defects because they should have had a vested interest in this topic.

The antenatal mothers group was expected to perform better than the general pool of postnatal mothers as they have had a dedicated time of counselling by a trained nurse/ counsellor.

From this study, the antenatal mothers unfortunately did not perform any better than the postnatal mothers (Table I). This can only be attributed to the fact that some of the antenatal mothers did not comprehend what they were counselled or simply did not remember the counselling given. Perhaps the postnatal mothers after going through the course of their pregnancy have acquired a considerable knowledge.

As more than 90% of pregnancies with structural abnormalities occur in mothers with no apparent risk factors, ultrasound screening must be made universally available. It was comforting to note that most of the mothers of both groups surveyed felt that an ultrasound scan was a necessary procedure[7].

The risk of chromosomally abnormal pregnancies with its associated structural problems increases with maternal age and the age specific risks have been worked out in a large study for DS[9]. While the age specific risks are not necessarily common knowledge, it must be appreciated that an elderly mother, regardless whether she is a primigravida or multigravida, will be exposed to similar risks and they should be given similar genetic counselling.

Most mothers surveyed wanted to know if their foetus was normal and would have opted to terminate the pregnancy if a severe or lethal malformation was detected. This view is shared in similar studies done in other countries[10,11]. Antenatal care should ideally be started in the first trimester for optimal management to be planned (Table IX).

It is now evident that management of a pregnancy should

start as soon as the pregnancy is confirmed. Ultrasound scanning plays an important role in antenatal care in diagnosing viability, gestation and identifying foetal abnormalities. Public awareness of the various abnormalities and the various facilities available that can identify these problems must be increased. It is a major task that the medical community has to take on in order to decrease the overall incidence of birth defects and the psychological trauma associated with it[12].

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