BIRTH DEFECTS – THE STATE OF AWARENESS

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ABSTRACT

As doctors and nurses are the primary sources of medical information, a simple survey was conducted in 1992 among doctors and nurses to determine their level of awareness of the incidence of and risk of having an abnormal as well as their knowledge of the use of ultrasound in pregnancy.

Only 10% of doctors and 23% of nurses were aware that the general risk of having an abnormal baby is greater than 1%. Only 37% and 17% of doctors and nurses respectively were aware that the best time to screen for structural abnormalities was indeed at approximately 20 weeks gestation.

Keywords: structural defects, chromosomal defects, ultrasound, education

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INTRODUCTION

In Singapore, like in all developed nations, the primary cause of perinatal mortality is undoubtedly congenital abnormalities. However, there has not been very much public education in this area and it was felt subjectively that awareness of this matter was very low. Thus this present survey was carried out within the Singapore General Hospital (SGH) to determine various individuals' perception of this problem and how their knowledge on this subject was obtained.

METHODOLOGY

A questionnaire (Fig 1) was designed to quiz different groups within SGH on their awareness of a woman's risk of having a child with either Down Syndrome or structural birth defects as well as their knowledge of the currently available prenatal diagnostic procedures. Questions as to what each individual perceived as adequate antenatal care was also asked.

Fifty-nine general practitioners and ninety-two nursing staff were sampled in this study. They were each asked to answer the multiple choice quiz.

SURVEY FINDINGS

General Risk

Table I – Perceived general risk of having an abnormal baby

Risk	GPs	Nurses
1 in 100 or more	0.2%	22.8%
1 in 500	22.0%	10.9%
1 in 1,000	39.0%	31.5%
1 in 10,000	27.2%	25.0%

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The survey revealed that many medical personnel, general practitioners (GPs) and nurses held the erroneous belief that the general risk of having an abnormal baby was less than or equal to 1 in 500. Only 22.8% nurses and 10.2% GPs thought the risk was 1 in 100 or more which was the closest to the actual incidence of 2 to 6% foetal anomaly (major and minor included).

Down Syndrome

Only 18.5% nurses knew that Down Syndrome was actually Trisomy 21; 43% and 30% nurses knew that Down Syndrome children were mentally retarded and a result of a genetic disorders respectively. Only one nurse had never heard of Down Syndrome before.

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

Risk of DS with maternal age	GPs	Nurses
No change	0	5.4%
Increased	98.3%	89.1%
Decreased	1.7%	3.3%

DS : Down Syndrome

Chromosomal Abnormalities

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

Risk with increasing parity	GPs	Nurses
No change	74.6%	67.4%
Increased	24.0%	22.8%
Decreased	0	5.4%

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 only 74.6% GPs and 67.4% nurses. Twenty-four percent GPs and 22.8% nurses thought that the risk actually increased, while 5.4% nurses thought that the risk declined.

Structural Abnormalities

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

Risk with increase in maternal age	GPs	Nurses
No change	47.0%	37.0%
Increased	53.0%	59.0%
Decreased	0	2.0%

As the risk of chromosomal abnormalities increases with maternal age, the incidence of their associated structural abnormalities will also increase, although the absolute incidence of structural abnormalities which occur de novo as a result of multifactorial inheritance does not change with age, the overall incidence of structural abnormalities will increase with maternal age.

Only 53% GPs and 59% nurses thought correctly that the risk of having a structurally abnormal child was increased with increasing age.

Table V – Perceived risk of having structurally abnormal babies with increasing parity

Risk with increasing parity	GPs	Nurses
No change	83.1%	74.0%
Increased	11.9%	16.0%
Decreased	0	1.0%
No response	5.0%	9.0%

83.1% GPs and 74% nurses thought that the risk of having an abnormal child did not change with increasing parity. While 11.9% GPs and 16% nurses thought that there was an increased risk with increasing parity. There was 5% and 9% of nonresponders to this question from the GPs and nurses groups respectively.

Use of ultrasound in pregnancy

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

Ultrasound is a necessity?	GPs	Nurses
Yes	51.0%	82.0%
No	49.0%	16.0%

Only 51% of GPs and 82% of nurses thought that ultrasound scanning was a necessary test in every pregnancy.

Perceived detection rate of foetal abnormalities on ultrasound

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

Rate	50%	60%	70%	80%	90%	100%
GPs	27.1%	6.8%	15.3%	22.0%	23.7%	3.4%
Nurses	22.8%	17.4%	14.1%	20.8%	15.2%	5.4%

Realistically, the detection of foetal abnormalities depends on many variables. However under ideal circumstances one can realistically expect to detect 75% of foetal abnormalities.

At one end, 27.1% GPs and 22.8% nurses thought the ultrasound scan could only detect less than 50% of the major abnormalities while at the other 25% GPs and 20.7% nurses expected the ultrasound scan to detect 90 to 100% of all abnormalities.

Timing of ultrasound scans in pregnancy

Table VIII – Perceived optimal time for a structural screening ultrasound scan of the foetus

Gestation	GPs	Nurses
12 weeks +	10%	48%
16 weeks +	44%	28%
20 weeks +	37%	17%
24 weeks +	2%	3%
> 28 weeks	2%	1%
Not necessary	0%	0%

Only 37% of GPs and 17% of nurses thought the best time to check on the normality of a foetus was at 20 weeks.

Gestation at which first visit was thought best

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

Gestation	6/52	12/52-20/52	20/52-28/52	28/52-36/52	> 36/52
GPs	76%	22%	–	–	–
Nurses	66%	30%	1%	–	–

Only 76% of GPs and 66% of nurses thought that the first visit to a doctor should occur two weeks after missing the expected menses. Twenty-two percent of GPs and 30% nurses felt that the first consultation could wait till some time between the third and fourth month of gestation. None of the GPs and only one nurse felt that the best time was between the fifth to the sixth month of gestation.

DISCUSSION

In recent years, foetal abnormality has emerged as the major cause of perinatal mortality as well as a significant contributing factor 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]. In an unpublished review of all the foetal malformations delivered in the SGH between January 1989 to April 1990, the incidence was also 1.53%. A review of antenatal diagnosis of foetal abnormalities by ultrasound between 1978 to 1980 in the National University Hospital (NUH) found that 20.7% of perinatal deaths were due to lethal malformations[2]. In SGH, congenital malformations have been the single largest contributor to perinatal mortality, ranging between 45% to 55% over the past four years.

The medical fraternity thus plays an important role in educating the public with regard to their risk of having either a chromosomally or structurally abnormal pregnancy. GPs and nursing staff constitute the two major groups within the medical profession that provide primary health care and education to the public.

Unfortunately, from our pilot study, it was noticed that a large proportion of these two groups of medical personnel are ill-formed with regard to the risks of both chromosomal and structural abnormalities in pregnancy.

This lack in information is further exemplified by their uncertainty of the usefulness of ultrasound scanning in both early and midtrimester pregnancy.

At present the common practice in most advanced medical health centres is to provide ultrasound scans for pregnancies early in the first trimester to exclude conditions like multiple pregnancies, ectopic pregnancies which are potentially fatal as well as to obtain an accurate dating in viable pregnancies[3].

A screening scan is then performed at around 20 weeks of gestation to exclude gross foetal structural abnormalities. This gestation is considered as the optimum time for screening as the anatomical structures are fairly developed and are usually well demonstrated on scan and most major or lethal foetal anomalies can be excluded by the trained eye[4,5]. In the event that a gross malformation or fatal anomaly is seen, selective termination is still a viable option for the parents as the pregnancy is still within the gestational limits of legal termination[6].

A detailed anatomical screening ultrasound scan can detect approximately 70-80% of gross foetal anomalies[7,8]. The anomalies which are not detected are often those which do not lead to severe morbidity or mortality or usually are amenable to postnatal correction. However, certain abnormalities may manifest themselves only in the third trimester eg microcephaly, intra-uterine growth retardation and as such a third trimester scan (at 32 weeks of gestation) provides a good opportunity to monitor the growth of the foetus as well as to identify anomalies which may not have been evident before.

More than 90% of pregnancies with structural abnormalities occur in mothers with no apparent risk factors as a result of multifactorial inheritance or mutations. As such, ultrasound screening must be made universally available to all mothers[9].

The risk of chromosomally and structurally abnormal pregnancies with its associated structural problems increase with maternal age and age specific risks have been worked out in large studies for Down Syndrome eg the Ferguson Smith's study which involved more than 50,000 pregnancies and has often been quoted as a standard[10]. 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 the appropriate genetic counselling.

In order that this ideal practice can be carried out, pregnant mothers must be seen early in pregnancy. A shared antenatal programme between the obstetrician and the general practitioner would be the most satisfactory arrangement allowing the patients the convenience of being followed-up by her own general practitioner while not missing out on the essentials of specialist obstetric care.

Educating all medical personnel would be the first step towards improving the public's awareness of the birth risks of both chromosomal and structural anomalies. They can then in turn disseminate accurate information to the pregnant mothers and direct them to centres which can provide them with the necessary prenatal diagnostic services.

CONCLUSION

The task at hand is now obvious, there is an urgent need to provide all doctors and nursing staff with an update as to the risks of foetal chromosomal and structural abnormalities. They must also be informed of the important role of universal ultrasound screening in pregnancy and the present state of the art in prenatal diagnosis. The need is even more pressing as the age of our primigravidae is increasing and more women are now contemplating larger families in their late thirties in line with the government's policies.

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Fig 1 – Birth defects survey questionnaire

1. What do you think is the general risk of having an abnormal baby?
   a) 1 out of every 100 or more
   b) 1 out of every 500
   c) 1 out of every 1,000
   d) 1 out of every 10,000
2. Do you think the risk of having a Down Syndrome birth changes with a mother's age?
   a) the risk does not change with age
   b) the risk decreases with age
   c) the risk increases with age
3. Do you think the risk of having babies with chromosomal abnormalities changes with the increasing number of children a woman has?
   a) the risk does not change
   b) the risk decreases with increasing number of children
   c) the risk increases with increasing number of children
4. Do you think an ultrasound scan is a necessary test for every pregnant woman? Yes ____ No ____
5. Do you think the risk of having babies with structural abnormalities changes with a mother's age?
   a) the risk does not change with age
   b) the risk decreases with age
   c) the risk increases with age
6. Do you think the risk of having babies with structural abnormalities changes with the increasing number of children a woman has?
   a) the risk does not change
   b) the risk decreases with increasing number of children
   c) the risk increases with increasing number of children
7. Babies with major abnormalities may be detected by ultrasound. I expect _____ to be picked up on scanning
   a) 50% or less
   b) 60%
   c) 70%
   d) 80%
   c) 90%
   f) 100%
8. When do you think is the best time to get an ultrasound scan in pregnancy to check on how normal your baby is?
   a) 12 weeks
   b) 16 weeks
   c) 20 weeks
   d) 24 weeks
   e) after 28 weeks
   f) not necessary
9. When do you think is the best time for first consultation of pregnancy?
   a) 2 weeks after missing expected period
   b) between 3 to 4 months
   c) between 5 to 6 months
   d) between 7 to 8 months
   e) at the beginning of the 9th month

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