CLINICAL MARKERS IN THE PRENATAL SCREENING OF CONGENITAL BIRTH DEFECTS: SIGNIFICANCE IN THE DEVELOPING WORLD

(A Prospective Study of 70 cases)

By

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SUMMARY

Seventy cases of congenital birth defects were studied to determine the applicability of clinical markers in their prenatal screening. A statistically significant 88.7% cases bore clinical pointers suggesting of their value in selecting patients for further in-depth investigations.

Introduction and Purpose

"My womb pregnant by thee,
and now excessive grown;
prodigious movement felt
and rueful throes;
at last this odious offspring
whom thou seest;
tore through my entrails....distorted!"

'Paradise Lost'—Milton

The most traumatic experience for a gravid woman, her husband and their family, is undoubtedly the unheralded birth of a deformed child, precipitating feelings of horror, depression, inadequacy and failure in the parents.

Major malformations occur in approximately 2.5 per cent of new born babies and are a leading cause of pregnancy wastage and neonatal mortality (King *et al*, 1982). Fetus surveillance techniques developed in the last decade now permit the prenatal diagnosis of many malformation (Rayburn and Barr, 1985).

But alas! We in the developing world stand disadvantaged. While we are greatly impressed by the blitz of specialised investigations emanating from the developed world, we tend to forget that our overburdened medical system forces us to offer the relevant facilities to but a fraction of our antenatal patients. What we need is a simple system, which would identify a small select group of patients at risk, for further in-depth biophysical and biochemical investigation, by which antenatal diagnosis is now possible (Donald, 1983).

In this paper we study the applicability of using antenatal clinical markers in the third trimester in screening out pregnancies at risk of potentially involving a malformed fetus.

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Materials, Methods and Parameters

Seventy cases of congenital birth defects out of a total 9960 deliveries were studied between 1st February, 1985 and 31st January, 1986—a study period of twelve calendar months.

Study Groups Sixty-two booked cases with birth defects where antenatal records were available comprised the group under study.

Control Group: One hundred randomly selected booked cases were studied to determine the general incidence of the clinical markers.

Clinical Markers

A history of previous fetal wastage and four objective antenatal clinical markers were selected for our study.

1. Non-engaged presenting part at and after 36 weeks of gestation.
2. Abnormal lie and presentations at and after 32 weeks of gestation.
3. Polyhydramnios.
4. Intrauterine growth retardation.

Analysis of statistical significance were performed by the Chi Square test, with $p > 0.01$ level highly significant.

Results and Analysis

A total of 70 cases with major congenital malformations were recorded giving an incidence of 0.7%.

The role of the antenatal clinic in Prenatal Screening

Antenatal follow-up and the detection of birth defects

At 2 or 3 visits 39.1 per cent cases were investigated, 30.4 per cent being diagnosed while at more than 4 visits 70.4 per cent cases were investigated, 40.7 per cent being diagnosed, reflecting the value of regular and repeated antenatal follow-up in detection of these cases.

The importance of history taking

Previous history of Reproductive Wastage

TABLE II

Previous History of Reproductive Wastage

No. of fetal losses	1	2 or more
Cases	10	5
Percentage of multigravidas	26.3%	13.2%

Table II highlights the importance of Reproductive wastage in the obstetric history of multigravidas in screening out cases at risk. 16.3 per cent of multigravidas gave a positive history.

3 multigravidas (7.9 per cent), gave a history of previous malformed babies.

Clinical Markers

1. *Non-engagement*

Non-engagement was detected clinically by the third and fourth manoeuvers of Leopold and Sporlin.[4]

Incidence of Non-engagement at and after 36 weeks

That 70.8 per cent of primigravidas in the study group had a non-engaged presenting part at and after 30 weeks, as compared to 30 per cent of cases in the control group is statistically significant from Table III.

TABLE I

Antenatal Follow up and the Detection of Birth Defects

No. of visits	1 visit	2/3 visits	4 visits/more
Total cases	12	23	27
Cases investigated	3	9	19
Percentage	25%	39.1%	70.4%
Cases diagnosed	3	7	11
Percentage	25%	30.4%	40.7%

TABLE III
Incidence of Non-engagement at and After 36 Weeks

	Total	Cases	Percentage
Primigravidas			
Study Group	24	17	70.8
Control Group	50	15	30.0
Highly significant, $p > 0.01$ level			
Multigravidas			
Study Group	38	28	73.7
Control Group	50	29	58.0
Total			
Study Group	62	45	72.6
Control Group	100	44	44.0

Neural Tube defect was the commonest cause of non-engagement.

2. *Abnormal lye and presentations*

Incidence of abnormal presentations at and after 32 weeks

As per Table IV the incidence of abnormal presentations was 45.2 per cent in the study group, as against the 16 per cent in the control group, a statistically significant finding. The Neural Tube defects were mainly found in cases with podalic presentations while the abdominal wall defects were associated more with transverse and oblique lyes.

3. *Polyhydramnios*

Incidence of Polyhydramnios

Table V shows that Polyhydramnios is probably the single most important clinical marker as evidenced by the 33.9 per cent incidence in the study group as against the 2 per cent incidence in the control group. The diagnosis in these cases was clinical, confirmed later by ultrasonography.

TABLE IV
Incidence of Abnormal Presentations at and After 32 Weeks

	Total	Cases	Percentage
Study Group	62	28	45.2
Control Group	100	16	16.0

Highly significant, $p > 0.01$ level

TABLE V
Incidence of Polyhydramnios

	Total	Cases	Percentage
Study Group	62	21	33.9
Control Group	100	2	2.0

Highly significant $p > 0.01$ level

4. *Intrauterine Growth Retardation*

As per Table VI intrauterine growth retardation was diagnosed in 14.5 per cent cases in the study group and confirmed by ultrasound.

5. *Fetal Bradycardia*

Three patients presented during pregnancy with a history of diminished fetal movements. Clinically fetal bradycardia was recorded. Monitoring showed a baseline of 90-100 beats per minute and a silent pattern.

Overall Incidence of Clinical Markers

Comparative incidence of Markers in the study and control groups

As shown in Table VII it is highly significant that 88.7 per cent of cases bore clinical markers in the study group as compared to 51 per cent in the control group. 53.22 per cent of cases in the study group bore 2 or more clinical pointers as compared to only 11 per cent in the control group.

Discussion

A review of 18,155 newborn infants revealed 464 major malformations—2.6 per cent. (Holmes) The 0.7 per cent incidence of major malformations in our study is probably a result of natural incidence in our population. In the developing world we are faced with a number of problems inherent to the social fabric and our medical services impending effective detection.

The likelihood of a case being investigated and detected were in direct proportion to the number of antenatal visits made, stressing value of regular and repeated follow-up to allow optimum screening.

That almost all physical anomalies are detectable early and accurately by ultrasonography and its as yet safe record, make it the investigation of choice. The overload on our facilities allows us to

TABLE VI
Incidence of Intrauterine Growth Retardation

	Total	Cases	Percentage
Study Group	62	9	14.5
Control Group	100	2	2.0

TABLE VII
Comparative Incidence of Markers in the Study and Control Groups

	Study Group (Total 62 cases)		Control Group (Total 100 cases)	
	1 marker	2 or more markers	1 marker	2 or more markers
Number of Cases	55	33	51	11
Percentage	88.7%	53.22%	51%	11%

Highly significant, $p > 0.01$ level

scan barely 15 per cent of our antenatal patients by ultrasonography at our hospital. Hence the problem here is to recognise cases at risk by mass methods of screening which can be applied to all women in pregnancy.[3] And what better tool do we have here in the developing world than our own clinical acumen?

A history of first trimester teratogenic insults and a history of previous reproductive wastage or a previous malformed baby may help limit selection. Obstetricians have been long aware of antenatal pointers in a pregnancy involving a malformed fetus. While Bader (1936) demonstrated that non-engagement of the fetal presenting part does not always reflect disproportion, our study shows a significant role in the screening of birth defects, particularly in primigravidas. By 32 weeks of gestation the general incidence of abnormal presentations is 11.3 per cent against a significant 45.2 per cent incidence in the study group. The high statistical significance of all the findings relating to the clinical markers in the study group stress their importance and reflect their applicability in the prenatal screening of congenital defects.

The presence of clinical pointers may be variously explained by a deformed fetal anatomy leading to altered polarity, relative disproportion due to abnormal fetal size, excess amniotic fluid volume, inhibition of fetal swallowing and impaired development of the autonomic nervous system. In our study, 21 cases were detected by ultrasonography (33.9 per cent). In 2 cases the findings were corroborated by radiology and elevated alpha fetoprotein levels. Nine cases escaped detection by ultrasonography cautioning us against total dependence on any single procedure. A judicious mix of the various aids would give optimum results.

Value of Third Trimester detection—

Today we are years behind offering inutero repair of malformations, in fact late registration often precludes the possibility of early diagnosis and selective abortion. But we can offer antenatal parental counselling regarding this condition, prepare them psychologically and emotionally to face up to this experience and help cushion the grief response.

Conclusion

As evidenced by this study a judicious use of clinical markers could help in prenatal screening out of a select group of patients at high-risk, for further in-depth Biophysical and Biochemical investigation by which detection of these defects is now possible.

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