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SHORT COMMUNICATION

PREVALENCE RATE OF DOWN'S SYNDROME IN KARACHI RESIDENT WOMEN

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Prevalence rate of Down's syndrome in one thousand women of Karachi were evaluated. The preliminary screening showed that 80 out of 1000 women fall in this category based on MSAFP. Further tests were augmented such as hCG, ME along with MSAFP confirmed that 0.2% patients delivered babies with Down's syndrome.

INTRODUCTION

Pakistan is a developing country and has limited resources to cater the needs of its vast health care system. The majority of the population (about 75%) of this country lives in rural areas where very meager or no facility is available on antenatal monitoring. Even in large cities there is insufficient provision for delivery/maternity services and antenatal care. Considering the case of Karachi, where only about 50% of the women population can avail these services, therefore, essential screening data to draw meaningful conclusion on Down's syndrome is not available. Therefore diagnosis of Down's syndrome, the pregnant women above 40 years would be carefully monitored for present study.

The literature survey revealed that Down's syndrome is the commonest single cause of severe mental retardation in children. It is the result of trisomy 21 arising from non-dysjunction of chromosome in the ovum close to the time of conception. This is usually a random event. Throughout the world the frequency is approximately 1.3 per 1000 births (Cuckle *et al*., 1991). In 1984, Merkatz and colleagues described the association between trisome (18 and 21) and low levels of MSAFP in mid trimester. In 1987, Bogart and colleagues described the association between Down's syndrome and elevated levels of human chorionic gonadotropin (hCG). Measurement of hCG has proved to be the single-most efficient screening parameter. Wald *et al* (1988) has given classical format of a Down's syndrome screen in the triple test which includes AFP (α-fetoprotein), oestriol and hCG. Benscerraf *et al* (1987), Nicolaides *et al* (1992a,b) and Savoldelli *et al* (1993) have proposed that ultrasound may become an alternative to biochemistry and that Down's syndrome fetus may be detected by ultrasound. Sheng and Xue (1994) described the relation between prognosis of pregnancy and serum progesterone and increase of hCG. Kishida *et al* (1995) diagnosed premature rupture of membranes with an improved α-fetoprotein monoclonal antibody kit. McDuffie *et al* (1996) evaluated, a two years experience in a health maintenance organization, with mid trimester maternal serum screening with α-fetoprotein (AF), hCG, and unconjugated estriol (UE) as a serum for fetal Down's syndrome. Cusick *et al* (1996) predicted the pregnancy outcome from the degree of maternal serum α-fetoprotein elevation. Bradley *et al* (1997) studied the prevalence of undetectable low second trimester maternal serum unconjugated estriol levels and the association with increased perinatal morbidity or mortality in pregnancies at risk for placental sulfates deficiency. Takayasu (1997) observed that during pregnancy inhibin is secreted from placenta but inhibin in maternal blood was examined in relation to that in cord blood, birth weight, placental weight and estriol (E_3) in maternal blood. Wallace *et al* (1997) recommended that the inhibin-A has been useful new perinatal marker of Down's syndrome, significantly increasing detection rates. In the light of these observations a study has been undertaken to evaluate the Down's syndrome in Karachi female population.

MATERIALS AND METHODS

Apparatus
Eliza Test System
(Enzyme linked immunosorbent assay method)
Boehringer Mannheim, GmBH, Germany.
Micrus Ultrasound System
Model SSD-500, Aloka, Japan.
Automatic Blood Analyzer
Model 705, Hitachi, Japan.

One thousand patients were randomly selected from various hospitals and maternity homes of Karachi, namely JPMC (Jinnah Postgraduate Medical Centre), Civil Hospital, Abbasi Shaheed Hospital and other hospitals in Karachi. High risk pregnancies were separated from normal pregnancies by taking detailed history, conducting regular antenatal examination and by caring out all routine investigations.

Blood samples from pregnant women were collected in the morning by veinepuncture. The blood was drained into the sterilized glass containers containing a little amount of heparin or EDTA (anticoagulant solution). The ELIZA and automatic blood analyzers were employed and applied to various determinations (D'Angelo *et al*., 1995 and Penttila

et al., 1995). Patients had regular ultrasound examinations during antenatal visits.

RESULTS AND DISCUSSION

Table and figure show the number of patients falling in different risk factors of Down's syndrome. One thousand antenatal patients were randomly selected for study, out of these patients, age of 80 patients was above forty years which makes a major risk factor for Down's syndrome, the percentage of which is 8%. These 80 women were high risk for Down's syndrome. Therefore, they were subjected to various blood tests. Maternal serum α-fetoprotein levels was measured in 15 patients out of 80 which showed below normal levels of MSAFP, decreased levels of MASFP is one of the important signs of Down's syndrome. The percentage calculated out of 80 patients was 18.75%. Further screening as well as few more tests were performed on these 15 women. Their blood levels of human chorionic gonadotropin hormone (hCG) and unconjugated serum estriol hormone were measured. Two patients out of 15 showed high levels of hCG and low level of μE_3. When these were combined with MSAFP, hCG and μE_3, all together were 90% confirmatory for Down's syndrome. These 5 patients were scanned for Down's syndrome. On ultrasound screening 2 out of 5 patients showed confirmed signs of Down's syndrome i.e. thickened mucal folds, short femur length, hypoplastic middle phalanx of 5th digit. On delivery these 2 patients delivered babies suffering from Down's syndrome. It is concluded from the above study that the rate of prevalence is 0.2% out of 1000 patients.

Table
Prevalence rate of Down's Syndrome
in Karachi resident women

S. No.	Risk factors	Number of patients (n)	Percentage
1.	Maternal age >40 years	80 (1000)	8.00
2.	MSAFP (>level)	15 (80)	18.75
3.	hCG (High) level μE_3 (Low) level	5 (15)	33.33
4.	Ultrasound scanning shows Down's syndrome	2(5)	40.00
5.	Babies with Down's syndrome on birth	2 (1000)	0.20

n = (Total number of patients).

Consequently our study shows that 0.2% cases out of 1000 birth is very low as compared to 1.3% of out of 1000 birth described by Cuckle *et al* (1991). The reason for this decrease in percentage is two to three folds. Firstly is due to the inaccurate collection of data because the turn up for antenatal monitoring is very less; secondly the early marriages wherein very few mother above 40 years were seen, and thirdly medical facility is not available for low income women.

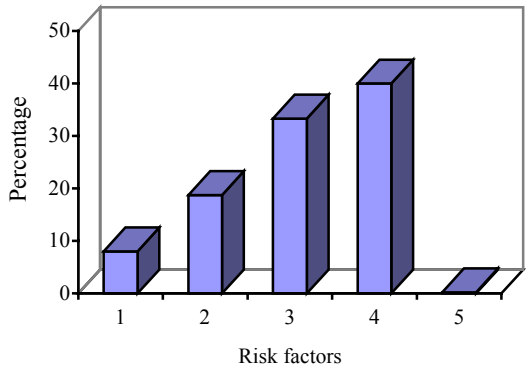

Fig.1: Prevalence rate of Down's Syndrome in Karachi resident women.

1. Maternal age > 40 years 80(1:000)
2. MSAFP level decreases 15(80)
3. hCG level high and μE_3 level low 5(15)
4. Ultrasound scanning shows Down's syndrome 2(5)
5. Babies showing sign of Down's syndrome on birth 2(1000).

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