health technology assessment

EXECUTIVE SUMMARY

MATERNAL SCREENING FOR FOETAL ABNORMALITY

HEALTH TECHNOLOGY ASSESSMENT UNIT
MEDICAL DEVELOPMENT DIVISION
MINISTRY OF HEALTH
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EXECUTIVE SUMMARY

INTRODUCTION

Congenital malformations are structural or anatomical defects that are present at birth, resulting from influences acting on the developing embryo in early pregnancy. Some congenital malformations are potentially preventable; however, they remain major causes of early death, hospitalization of infants and young children and significant long-term physical and developmental disabilities. Screening and early detection of Downs Syndrome and other chromosomal anomalies in-utero provides several benefits like the opportunity to inform parents and counseling on the likelihood of delivery of an affected baby. This would allow them subsequently to make an informed decision about whether to continue with the pregnancy or alternatively undergo selective therapeutic abortion to avoid the birth of a disabled baby. The other potential benefits of maternal screening and early diagnosis include preparing the parents psychologically for the delivery of a handicapped baby, enabling doctors to better prepare for the delivery and care of baby, and also avoidance of unnecessary caesarian section in cases with lethal chromosomal abnormality. In addition, maternal screening can also identify foetuses with open neural tube defects.

OBJECTIVE

The objective of this assessment is to determine the effectiveness, safety, cost implications, ethical, legal and social implications of maternal screening for foetal abnormality of the following conditions: Downs Syndrome, Neural Tube Defects and Thalassaemia.

RESULTS

(1) Downs Syndrome

A literature review found that screening using triple serum markers during the second trimester combined with ultrasound to date gestation, increased the detection rate of Downs Syndrome compared to the use of the last menstrual period, so as to avoid performing amniocentesis and chorionic villi sampling (CVS). Ultrasound is used as a secondary tool if serum marker results are positive. It also found that thickened nuchal fold visualized by ultrasound in the second trimester is not a practical screening tool for decisions on offering amniocentesis. However screening using serum markers followed by invasive prenatal diagnosis by amniocentesis or CVS has to be carried out with caution. It was also found that a screening programme is more cost effective than caring for Downs Syndrome children. There are serious ethical and religious issues in screening of maternal serum. It can be concluded that there is sufficient evidence to support the use of triple serum markers combined with ultrasound in second trimester, but there are major ethical and religious issues especially among the Muslim population.

(2) Neural Tube Defects

There is evidence that the use of maternal serum alpha- fetoprotein and ultrasound is effective to detect neural tube defects. However, there is insufficient evidence to support the use of other modalities of screening. It was found that amniocentesis might cause spontaneous abortion. A

screening programme showed cost benefits. However, there are major ethical and religious issues especially among the Muslim population.

(3) Thalassaemia

A literature review found that screening tests like MCV/Red cell indices; Hemoglobin A2 estimation, Hb electrophoresis using high performance liquid chromatography or isoelectric focusing; DNA mutation analysis; polymerase chain reaction and other modalities like anti-zeta antibody test are the effective modalities for screening for thalassaemia, However, again there are major ethical and religious issues especially among the Muslim population.

RECOMMENDATION

Due to the ethical and religious issues surrounding prenatal screening, invasive diagnostic procedures and termination of pregnancies, a national programme of routine antenatal maternal serum screening for Downs Syndrome, neural tube defects and Thalassaemia is not recommended. However, screening should be made available to women who request for the test.