



Down Syndrome Prenatal Testing and Counseling

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Down Syndrome

- 1 in 800 live births
- Most common chromosomal anomaly
- Risk increases with maternal age > 35
- Majority of infants with Down syndrome are born to women < 35 due to the greater number of births in this age group
- 40% - 55% of infants with Down syndrome have cardiac defects
- Down syndrome is associated with gastrointestinal disorders
- Survival has increased to 90% at 5 years of age
- Down syndrome, in isolation, has a 99% survival rate at 5 years of age
- The life span of individuals with Down syndrome has dramatically increased



Prenatal Testing and Counseling

- American College of Obstetricians and Gynecologists recommends screening offered to all women regardless of maternal age
- Screening Tests
 - Maternal Serum Screening
 - Nuchal Translucency
- Diagnostic Tests
 - Chorionic Villi Sampling
 - Amniocentesis
- 85% - 95% of prenatally diagnosed Down syndrome pregnancies are terminated
- Non-directive counseling is considered the standard practice



Maternal and Child Health Concerns

- Prenatal testing is frequently perceived as routine prenatal care rather than simply an option
- Test uptake variation can be influenced by the knowledge and attitudes of the person providing the information
- 80% percent of genetics professionals stated they would terminate their pregnancy if they received a diagnosis of Down syndrome
- Physicians have lower expectations of people with intellectual disabilities than do other professionals
- Only 66% of women reported they had been counseled regarding both options, despite non-directive counseling being the standard
- Informed consent centers around risks and benefits of the procedure as opposed to the other implications which many women may not consider
- Support groups and contact with parents of children with Down syndrome are underused
- Low socioeconomic status does not increase risk of Down syndrome, yet in the United States it is associated with decreased prenatal testing and increased prevalence of Down syndrome live births

Recommendations

- Healthcare providers should reflect on their knowledge, attitudes, and beliefs
- It should not be assumed that women undergoing testing wish to pursue termination upon a positive test result
- Improve societal attitudes toward people with intellectual disabilities
- Further research is needed to better understand the knowledge, attitudes, and beliefs of those providing testing and counseling regarding people with Down syndrome, develop effective and comprehensive counseling standards, and eliminate disparities in testing.

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