The Development of Prenatal Diagnostic Tools
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Introduction

- What limitations currently exist to regulate the prenatal genetic diagnostic tools, and what limitations should exist if at all?
- Common Prenatal Genetic Diagnostic Tools currently used:
  - Amniocentesis
  - Chorionic Villus Sampling
  - Fetal blood sampling
  - Pre-implantation genetic diagnosis with in vitro fertilization
  - Post-natal genetic diagnosis

Where are all the Girls in China?

- In 1980, a one-child policy was imposed to slow the rapid population increase
- Cultural preferences for male heirs remained strong, and illegal prenatal sex selection has resulted in a gender imbalance
- The male-female ratio has risen to 130 boys for 100 girls
- Census in 2005 revealed that for those under the age of 20, males exceeded females by 35 million
- Consequences: Societal implications where there is a lack of females for men to marry

Significance of Cystic Fibrosis (CF)

- Autosomal recessive genetic disorder
- Caused by a mutation CFTR gene on chromosome 7
- Most common cause of chronic lung disease in children and adults, and most common fatal genetic disorder in caucasians
- Symptoms: Severe lung damage and nutritional deficiencies, failure to grow, and respiratory infections, death by age 2 without treatment
- Prevalent in those with Eastern European lineage and in Ashkenazi Jewish communities
- Carrier testing in those with a family history of CF is encouraged though the test is expensive
- Diagnostic test for infants is a Sweat Test, measuring the amount of sodium and chloride in the sweat

Prevalence of Carrier Testing:

- Currently, just those with a family background of CF are tested if they are carriers
- Jews of Eastern European descent are regularly tested regardless of family history

Conclusion

- In various cases, mandating or strong cultural advocacy towards carrier testing has almost entirely eliminated certain genetic disorders from some populations
- Providing options earlier for families when a risk exists for a genetic disorder will decrease cultural, emotional, and financial burdens in the future
  - However…
- Should carrier screening be mandated or just highly encouraged?
- Should carrier screening be increased for cystic fibrosis?
- What is the role of carrier screening and prenatal genetic screening for genetic diseases?

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