Who is Affected by Genetics?

Genetics is a rapidly expanding field that influences the practice of medicine throughout the lifecycle from before conception through adulthood. This field addresses hereditary disorders, birth defects, and complex diseases where both genetic and non-genetic factors play a role in the development of a disorder. Although the single gene disorders are rare, collectively they comprise over 15,500 recognized genetic disorders and affect 13 million Americans (Doyle 1).[12] A single gene disorder results when a mutation causes the product of a single gene to be altered or missing such as sickle cell disease or phenylketonuria (PKU). Although this figure is significant, the broader view of "genomic" medicine will provide opportunities for health promotion and preventive medicine throughout the lifecycle. Genomics is a new term whose standard definition is evolving; however, the Centers for Disease Control and Prevention (CDC) use the term genomics to refer to "new information emanating from the Human Genome Project" (*FAQ's* 2).[18] Advances from the Human Genome Project promise to revolutionize the practice of medicine. Just as immunizations (from polio to influenza vaccinations) are universally accepted as effective preventive medicine throughout the lifecycle, genetics will soon be considered an essential component of preventive medicine for all Americans. Dr. Victor McKusick reports genomic medicine will render medicine "more predictive and, therefore more preventive" (2294).[33] Comprehensive "genome screens" for recognition of an individual's susceptibility to common disorders can be foreseen. Clinical diagnosis will become more specific and precise, and treatments more specific and safer (McKusick 2294).[34] Genomics will provide opportunities for the individualization of medical care to achieve the "right treatment for the right patient" (McKusick 2294).[35] Better understanding of an individual's genomic make-up should permit drug therapy to get away from the one-size-fits-all approach. It should allow the selection of drugs to be more effective in the treatment of a given disorder in a given individual; thus improving treatment outcomes and decreasing the risk for adverse side effects. The development of new genomic-based drugs are expected to produce completely new lines of medications for disorders that are currently not treatable, and provide more effective and safer alternatives for drugs that are currently in use today (McKusick 2294).[36] Genomic medicine will enhance medical care to include presymptomatic identification of susceptibility to disease, preventive interventions, selection of pharmocotherapy, and individual design of medical care based on genotype. Genomic medicine promises to improve and promote the health of all citizens.

The Stottlemyre Family meets with a geneticist for diagnosis and management of a common genetic disorder in their family.

[12] *Doyle, Debra, ed. . .*
[18] *FAQ's About Workforce. . .*
[33] *The Anatomy of Human Genome...*
[34] *The Anatomy of Human Genome...*
[35] *The Anatomy of Human Genome...*
[36] *The Anatomy of Human Genome...*

Genetics Throughout the Lifecycle

Stage of Life		Genetic Service
Preconception		A couple receives genetic counseling regarding their risk for having a child with sickle cell disease. Also discussed are ways to decrease the risk for birth defects (such as folic acid to reduce the chance of neural tube defects, and that smoking has been related to a higher chance of prematurity and low birth weight). For optimal birth outcome, every couple should have preconception counseling and genetic testing offered as indicated.
Prenatal		A pregnant women gives blood to screen for birth defects. Every woman should be offered appropriate genetic counseling and testing. Offering screening for neural tube defects and Downs syndrome or determining carrier status for genetic disorders such as sickle cell disease or cystic fibrosis is standard medical practice.
Newborn		A newborn has been identified through newborn screening with the genetic disorder phenylketonuria (PKU). PKU is difficult to detect without a blood test and if left untreated can result in profound mental retardation. Newborn screening prevents mental retardation through early identification and treatment. In Oklahoma, every newborn is required to be screened for the genetic disorders of PKU, galactosemia, sickle cell disease, and for congenital hypothyroidism.
Child		A family receives genetic counseling about their child's risk for neurofibromatosis (NF) and recommendations for medical management for affected family members. Families with NF are referred to a geneticist for diagnosis and multi-disciplinary management for optimal health outcomes.
Adult		A women receives genetic counseling regarding her risk for breast cancer and possible genetic testing. Key components of the counseling session will include: risk analysis, pros and cons of genetic testing, preventive treatment options such as prophylactic surgery or chemoprevention, and the implications of testing for the woman and her family.

United States Data on Genetics and Birth Defects Throughout the Lifecycle

Preconception/ Prenatal

- 1 in 10 women do not know folic acid can prevent birth defects.[1]
- 12% of pregnant women receive inadequate prenatal care.[1]
- 3.5% of women drank heavily during pregnancy.[2]
- 13% of women smoke during pregnancy.[3]

Infancy

- 3 to 5% of all newborns have congenital malformations.[4]
- 0.5% of all newborns have a chromosomal abnormality.[4]
- 7% of all stillborns have a chromosomal abnormality.[4]
- 20-30% of all infant deaths are due to genetic disorders.[5]
- 30-50% of post-neonatal deaths are due to congenital malformations.[6]
- Alcohol-related damage (i.e., mental retardation) in 50,000 infants could be prevented if pregnant women did not drink.[2]
- 4 million newborns are annually screened at birth for treatable genetic conditions.[7]
- 10% of infant deaths (estimated) could be prevented if pregnant women did not smoke.[3]

Children

- 11.1% of pediatric hospital admissions are children with genetic disorders & 18.5% are children with other congenital malformations.[8]
- 50% of mental retardation has a genetic basis.[9]

Adult

- 12% of adult hospital admissions are due to genetic causes.[9]
- 15% of all cancers have an inherited predisposition.[10]
- 10% of the chronic diseases (heart, diabetes, and arthritis) that occur in adults have a significant genetic component.[11]

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Oklahoma Data on Genetics and Birth Defects Throughout the Lifecycle

Preconception/Prenatal

- 64% of women reported that a physician talked to them before they got pregnant about how smoking could affect a pregnancy.[1]
- 64% of women reported that a physician talked to them before they got pregnant about how alcohol could affect a pregnancy.[1]
- 79% of women reported that they have heard or read that taking folic acid can prevent some birth defects.[1]
- 80% of women reported that a physician talked to them during a prenatal visit about how smoking during pregnancy could affect their baby.[1]
- 80% of women reported that a physician talked to them during a prenatal visit about how drinking alcohol during pregnancy could affect their baby.[1]
- 90% of women reported that a physician talked to them during a prenatal visit about the kinds of medicines that were safe to take during pregnancy.[1]
- 74% of women reported that a physician talked to them during a prenatal visit about how using illegal drugs could affect their baby.[1]
- 77% of women reported that a physician talked to them during a prenatal visit about doing tests to see if their baby had a birth defect or genetic disease.[1]

Infancy

- 100% of newborns (approximately 49,000) are annually screened at birth for treatable genetic conditions.[2]
- 3.7% of live births have a birth defect.[3]
- 0.2% of live births have a chromosomal abnormality.[3]
- 3.9% of stillbirths had a chromosomal abnormality.[3]
- 21.4% of infants who died at less than one year of age had a congenital anomaly.[3]

Children

- 4% of hospital discharges of children 19 years of age and younger are related to a genetic disease or birth defect.[4]

Adult

- 14.4% of hospital discharges of adults greater than 19 years of age are related to a genetic disease or birth defect.[4]

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2. Oklahoma State Department of Health Newborn Screening Program.
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