



Human Genome Project

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1. What is the Human Genome?



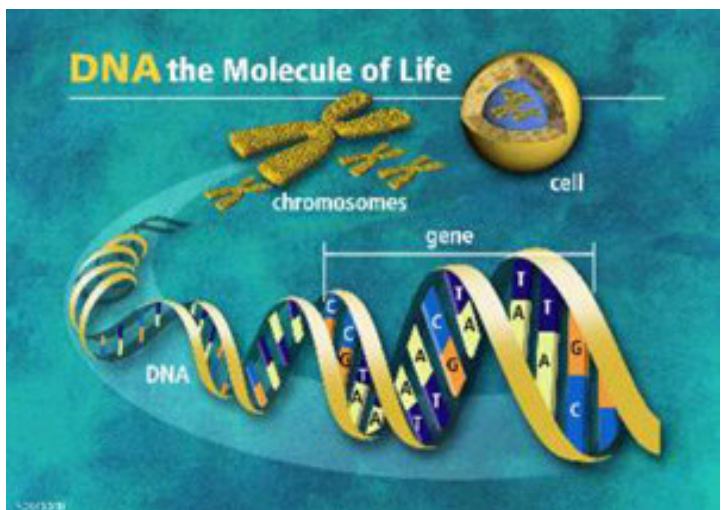
The Genome is the collection of all the genes contained in an organism. It is a chemical code, similar to a computer code that instructs a computer how to operate, and this code is found in each of the 100 trillion cells in your body. The human genome controls the manufacturing and building for the entire human body. All this information is contained in a cell at the very beginning of life, as soon as a sperm fertilizes an egg. This “one-cell zygote” divides and duplicates the entire genome into the two cells it produces. Those two cells divide and double and so on. This process continues and DNA remarkably controls the differentiation of cells into each of the 210 different varieties of tissues in the human body, from your teeth to your toenails and your corneas to your cardiac system. The entire genome is contained in every cell of an adult human’s body, but through a poorly understood control mechanism, the DNA only “expresses” part of the code in each cell, depending on the cell’s function.¹

How Does It Work?

1. Chromosomes: All the DNA in a single cell is on 46 chromosomes - 23 from the father and 23 from the mother (half from each). One of the chromosomes given from each parent is an “X” or “Y” chromosome. Women can only give “X” but men can give “X” or “Y”. If he gives an “X” chromosome, the child is a girl. If he passes down a “Y” chromosome, the child is a boy.

2. DNA: The chromosomes are made of DeoxyriboNucleic Acid (DNA). The DNA on all the chromosomes is about 2 meters, or 6 feet long and measures 50 trillionth of an inch wide. If you put all the DNA in your body end to end it would wrap around the globe a total of 4,734,848 times.

3. Nucleotides: DNA is a twisted ladder-like molecule. The side of the ladder is made of sugars and phosphates. The rungs of the ladder are made of four nitrogen compounds: adenine (A), thymine (T), cytosine (C), and guanine (G)]. One rung of the ladder with its side rails equals



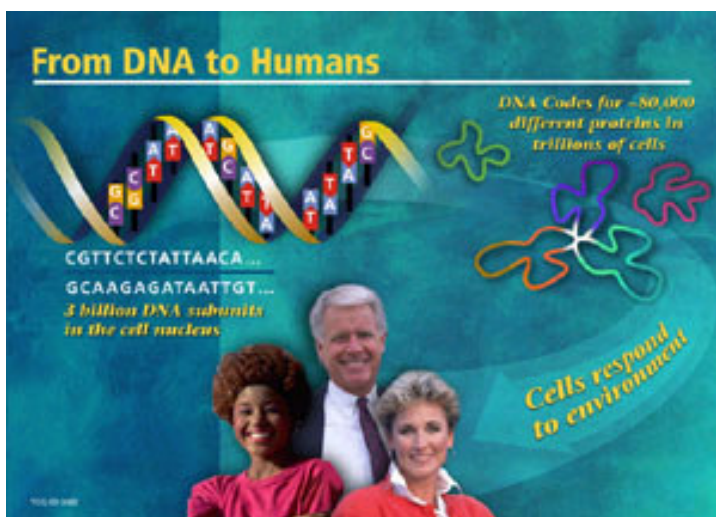
U.S. Department of Energy Human Genome Program

a nucleotide. Cytosine always pairs with guanine and thymine always pairs with adenine. This gives four possible letter codes (CG, GC, TA or AT), which are called base pairs.

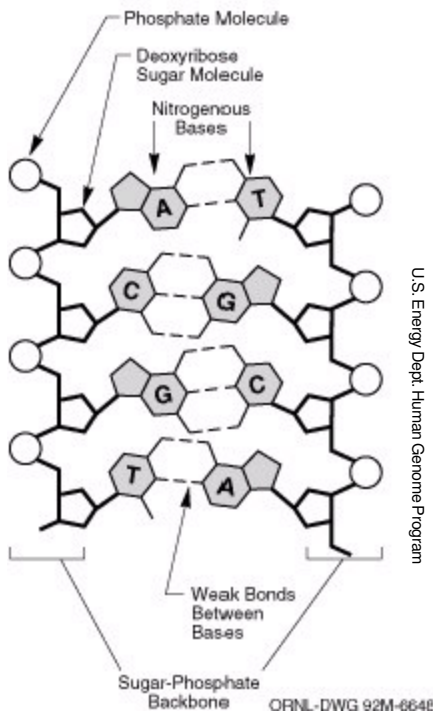
4. Proteins: The sequence of this “digital” code controls the production of amino acids which make up proteins, the basic building blocks for all the structure and function in the human body.

- There are only twenty amino acids.
- Three base pairs in the gene code for one amino acid.
- The proteins made are determined by the DNA in each cell.

Proteins make up your muscle, lungs, hormones, digestive enzymes and much more of what makes your body work. There are approximately 30-40,000 different proteins in the human body.



U.S. Dept. of Energy Human Genome Program



5. Genes

- An average of 3,000 base pairs make a gene.
- There are about 30 - 40,000 genes in the 46 chromosomes.
- Genes only make up 5 percent of chromosomes.
- The other 95 percent of DNA is part of the regulatory mechanism and/or has unknown functions.

All people are born with missing or damaged genes. That means you have 50-60 abnormal genes. Most people have no visible effects but some abnormal genes cause an increased risk of cancer, diabetes or other diseases. Some defects are passed down from parents; that is why some diseases run in families. For example, there is a gene that markedly increases the risk for colon cancer at a young age. Cystic Fibrosis (a defect of three base pairs that causes thick secretions resulting in recurrent pneumonia and early death) has a familial component. Other defects happen through mutations. When the cell divides to reproduce itself, it must replicate its DNA. The twisted ladders split forming two strands and an identical copy of the other side of the "ladder" is made. If a mistake is made, it can be passed on to subsequent cells. Some cancers and other genetic defects are caused in this manner. Other damage is due to radiation or other environmental injury to DNA.

2. What You Should Know



U.S. Energy Dept.
Human Genome
Program

What is the Human Genome Project?

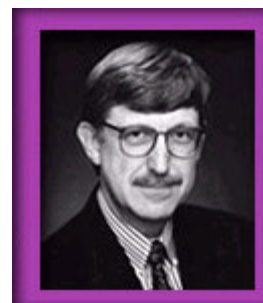
The U.S. Human Genome Project began in 1990 as a 13-year effort directed by the U.S. Department of Energy and the National Institutes of Health. Originally planned to last 15 years, technological advances have accelerated the expected completion date to 2003. Knowledge about the effects of DNA variations among individuals can lead to revolutionary new ways to diagnose, treat, and someday prevent the thousands of disorders that affect us. Besides providing clues to understanding human biology, learning about nonhuman organisms' DNA sequences can lead to an understanding of their natural capabilities that can be applied toward solving challenges in health care, energy sources, agriculture, and environmental cleanup.

Human Genome Project Goals:²

- Identify all the approximately 35,000 genes in human DNA
- Determine the sequences of the 3 billion nucleotide pairs that make up human DNA - this is essentially completed
- Store this information in databases
- Develop faster, more efficient sequencing technologies
- Develop tools for data analysis to understand the meaning of the genetic code
- Address the ethical, legal, and social issues (ELSI) that may arise from the project.

The National Human Genome Project was conceived by Charles DeLisi at the Department of Energy in October of 1985, but most scientists were skeptical that it needed to be done or actually could be done. After all, only 5 percent of DNA is actually genes, so why map all of it? And if the technology was even available, it was too expensive and would take too long. A team of leading scientists ultimately endorsed the concept in 1988—unanimously. Presently headed by Dr. Francis Collins, the project was funded with \$3 billion over 15 years and started in 1990 with the hope of reaching its goals by 2005. Since then, 18 countries have participated and over 200 principle investigators have been involved. Over \$2.5 billion had been spent in the U.S. on this project through the year 2000, with \$360 million spent that year alone. The Department of Energy has been involved in the human genome project because it was charged "with studying and analyzing genome structure, replication, damage, repair and the consequences of genetic mutations, especially those caused by radiation and chemical by-products of energy production."

Dr. Francis Collins



National Human Genome Project: Ahead of Schedule

Less than 3 percent of the human genome was mapped by May 1998. Celera Genomics, a private biotechnology firm, announced they would beat the US Human Genome Project and sequence the genome first. This, combined with improved mapping technology, accelerated the pace. By November 1999, 1 billion base pairs had been mapped. By June 2000, six months later, 2.7 billion pairs had been officially mapped, and the White House announced the 90 percent completion of the Human Genome Project. The significance of this feat was compared to landing on the moon, and soon the media was hinting that many diseases were very close to being cured. There were even reports that the genome map proved there was no need for a God. Mapping the human genome is similar in significance to finding a whole new unexplored continent. If the four-letter alphabet of 3 billion base pairs were printed out on computer paper, it would make a stack as high as the Washington Monument. Research ahead will focus on identifying genes, understanding control mechanisms and their relation to health and disease. The genome will be the foundation of scientific research for decades and affect each person's healthcare.

Potential Benefits of Mapping the Human Genome³

I. Molecular Medicine (Genetic Testing):

Improved diagnosis to and develop tests for many more diseases. There are already genetic tests for cystic fibrosis, phenylketonuria and a number of other diseases.

A. Earlier detection of genetic predisposition to disease. Doctors now must screen every patient as they get older for colon cancer. After age 50, women must undergo the discomfort and cost of a yearly mammogram. Genetic markers may allow physicians to identify patients with increased risk and screen them more frequently. Those patients with low risk may not need to be screened at all.

B. Rational drug design: If scientists understand the genetic cause of a disease, they may be able to design a drug that specifically blocks its expression.

1. For example, HER-2 is a receptor protein on breast cancer cells that promotes rapid growth of the tumor. Scientists were able to genetically engineer an antibody called "Herceptin" that blocked this receptor and thus markedly improved the survival of these women.

C. Pharmacogenomics: Through genetic screening scientists can understand the risk for side effects from a specific drug for a patient and give an alternate drug with fewer side effects.

D. Gene Therapy: The goal is to slip a corrective gene into a patient's DNA to replace a damaged or missing gene. The first successful case of gene therapy was reported in France, but unexpected complications have slowed research in this area.

II. Microbial Genomics

A. New Energy Sources: A "bio-fuel" may be made from bacteria modified to produce carbon fuels as a byproduct.

B. Environmental Monitoring: Pollutants may be detected by the discovery or design of microbes that are fluorescent or give other indications of oil or other pollutants in the environment.

C. Protection from Bioterrorism: Genetic design of vaccines to protect people from biological or chemical warfare; or scientists may be able to direct genetic modification to make people immune from biological agents.

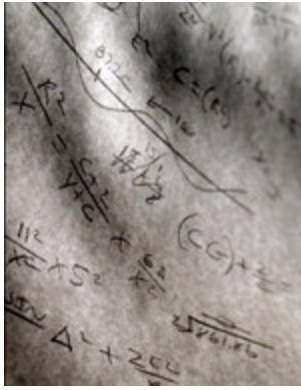
D. Safe Toxic Waste Cleanup: Non-pathological bacteria and other microbes can be developed to digest toxic waste and turn it into non-toxic materials.

III. Risk Assessment: The ability to better understand the risk of mutations from radiation, environmental toxins and drugs.

IV. Bioarcheology: Understanding the origins of people. Through genetic markers, scientists can determine what people groups are related or descended from others.

A. Example: Just as their ancestors believed, the Bantu-speaking people of South Africa, called the Lemba, were found to be descendants of Jews from Middle East approximately 2,700 years ago.

Other Benefits of the Human Genome Project: DNA Forensics



- **Identify criminals** from DNA in sperm, skin cells, saliva, blood or other biological materials left by criminal at scene of crime.
- **Exonerate the innocent:** Numerous prisoners have been released after new genetic tests on old biological materials showed they had not committed the crime they were accused of.
- **Identify cadavers:** Used extensively by military personnel who collect blood to get “genetic fingerprint.”
- **Paternity and family identification:** Scientists can tell if persons are genetically related when fatherhood or motherhood is unknown. Children could be identified years after they are kidnapped.

- **Organ donor match:** Although this has been used for a number of years, the process will allow more extensive markers of compatibility or incompatibility. As this improves, there will be fewer problems with tissue rejection after transplantation.
- **Agriculture & Animal Husbandry:** Scientists can produce more disease, insect and drought-resistant crops, healthier, more productive, disease-resistant farm animals, more nutritious produce and biopesticides. They will be able to design bacteria or viruses to kill or limit pests, and possibly design edible vaccines that could be incorporated into food products.

3. What You Can Do

Know the Social, Ethical & Legal Issues

“We are now in the Genomic Era. We must take seriously the possibility that some of this information may be used in less than benevolent ways.”

—Francis Collins, MD, National Human Genome Institute Dir.

Five percent of the National Human Genome Project—a total budget of \$125 million—went to study the social, ethical and legal issues of the project. As the first government project to have such a large source of funding, this portion of the Human Genome Project became the world’s largest bioethics program. Issues addressed include the following:



Privacy

A genetic profile absolutely identifies an individual. As the human genome is better understood, a person’s genetic ID will also reveal the risk for a multitude of diseases. Can confidentiality be maintained? Is confidentiality an absolute principle? What are the exceptions? For example, does a family member of a patient with a higher risk have a right to know that they have an increased risk of the disease as well?

- “One’s genetic information is the most personal and highly sensitive information one could possess. We can be separately identified from an unlimited number of persons through a DNA fingerprint.” —C. Ben Mitchell, *Genetic Engineering*⁴
- Genetic information will become more and more valuable to health and life insurance companies because it will reveal the risk for illness, disability and death. DNA samples can be held indefinitely and could be used as part of national identification database. The federal government already stores genetic the ID of military personnel and all convicted felons. A lot more will be gleaned from these stored samples ten years from now—information that the person may not want others to know.
- Confidentiality is a long-held principle of medicine and is contained in the Hippocratic Oath: “*And whatever I shall see or hear in the course of my profession,... if it be what should not be published abroad, I will never divulge, holding such things to be holy secrets.*”

- There is presently no federal legislation dealing with the issue of genetic privacy, except for President Clinton's executive order banning the use of genetic information in the hiring or promotion of federal employees on February 8, 2000.
- Genetics, Religion, and Ethics Project of the Institute of Religion Summary Statement: "*Religious values mandate the defense of personal privacy, integrity of the family and good social relations. Therefore, we support policies and methods of securing consent for access to genetic information obtained through screening. Moreover, the use of confidential information must be carefully circumscribed to avoid embarrassment, social stigmatization, disruption of marital and family relations, and economic discrimination. Care should be taken to avoid or prevent the unjust use of an individual's genetic data.*" — *Contemporary Issues in Bioethics*⁵

Discrimination

Genetic information has already allowed discrimination.

- Sickle cell screening of African Americans in the 1970's resulted in loss of health and life insurance and dismissal from military.⁶
- Thirty-two separate incidents were reported in New England of discrimination in health, life, disability, mortgage and auto insurance, as well as seven cases of discrimination in unemployment. Many of these people were still healthy but had undergone testing because of relatives with the disease.⁷
- A 1996 Stanford and Harvard study revealed 206 cases of discrimination by businesses, insurance agencies, schools, blood banks and the military in which women were persuaded or coerced to abort babies with the cystic fibrosis gene.⁸



Stigmatization

Stigmatization is internal discrimination when we think less of ourselves than we should. Many clinicians default to the position that children with genetic defects like Down's Syndrome should be aborted because the child is seen as having a life not worthy to be lived. Parents are often given the impression that they have no right to bring such a child into the world because it is a burden to society.

- People found to have the gene for Huntington's Disease, which always causes an unpleasant death in mid-life, have not only become anxious and depressed but some have committed suicide years before having any symptoms.
- Genetic counseling is mandatory for anyone considering genetic testing and further counseling is needed to interpret the results of their tests. Some patients may opt not to have testing because at present nothing can be done for many of the conditions identified, and a positive diagnosis may harm rather than help them.
- Not only is informed consent essential but there is also a risk of false positives (the test is positive but the person doesn't have the gene) and false negatives (the test is negative but the patient actually has the gene) in any genetic screening, even when the same sample is tested by different labs. Patients must have the opportunity to weigh the benefits versus the risks.
- Christians have a special obligation in this area to show the love of God to people with genetic diagnosis. We must value their lives and let them know that God does too no matter what genetic diagnosis they carry.
- Linus Pauling, advocate for Vitamin C suggested that "*there should be tattooed on the forehead of every young person a symbol showing possession of the sickle-cell gene or whatever other similar gene, such as the gene for phenylketonuria [PKU] If this were done, two young people carrying the same seriously defective gene... would recognize this situation at first sight and would refrain from falling in love with one another.*"



"I sat in horror as he relayed to me the details of the disease. He painted a bleak, hopeless picture and told me that I should consider having my tubes tied to avoid having children. He did not bother to refer me to any self-help groups, genetic counseling, or the Huntington Disease Society of America. The doctor left me feeling hopeless and of little value to myself or others. I was certain that I was damaged and defective and that no one would ever love me. Looking back, it was no wonder I sank into a depression which became progressively deeper from that day on, culminating several years later in two attempts to take my life." —Huntington Disease Patient⁹

Eugenics

Eugenics is defined as “the study of hereditary improvement of the human race by controlled selective breeding,”¹⁰ Darwin followers in the late 1800s thought eugenics would guarantee that “more suitable races would prevail.”¹¹

Positive Eugenics is used to facilitate breeding among those thought to have desired physical and intellectual characteristics, (i.e. to create the “master race”) while Negative Eugenics is used to discourage breeding among those with mental or physical disabilities. Forced sterilization of “defectives” became law in many states in the early 20th century. The first was passed in Indiana in 1907. By the 1920s, 28 states had followed Indiana’s lead and as a result, 15,000 individuals were forcibly sterilized. Germany followed with its sterilization laws in 1933. Over 400,000 people were forcibly sterilized between 1934 and 1937. This ultimately led to the euthanasia of 70,000 people with genetic defects or other incurable illnesses between 1939 and 1941.¹²

Today, there is a “new” eugenics movement. The new negative eugenics advocates and many states mandate prenatal screening to identify genetic defects or other malformations in unborn children. Doctors can be sued if they fail to offer prospective parents the option of aborting a child with Down’s syndrome and a growing list of other conditions. There is a new assumed “right” to reproduce and have a normal child that trumps any ethical concerns about any methodology such as surrogacy, egg or sperm donation and even human cloning.

Positive eugenics today advocates sex selection for “family balance” and selecting the “best” embryos for implantation after in-vitro fertilization. The next frontier is designing your children to through trait selection, genetic enhancements or even cloning.

In vitro fertilization specialists today create multiple embryos in the lab and then remove a cell from each developing embryo to test genetically so that the “best” embryos can be implanted. The rest are discarded. Sex selection techniques are also possible through semen separation.

If these practices became wide spread, they would have social consequences. A survey revealed that many couples would do sex selection if it was reliable and cheap. Ninety percent of couples would choose a boy first and 72 percent wanted a boy if they could have only one child.¹³

Human cloning, which is already taking place in private institutions, is the process of removing the nucleus from an adult cell and fusing it into an enucleated egg with an electrical charge. The cells quickly begin dividing to form an identical genetic

copy of the cloned person. Implantation of a cloned embryo and the subsequent delivery of the first human clone is thought to be inevitable.

Biological Determinism

Biological determinism is the concept that our genes control us like a computer program runs a computer. For example, the so-called homosexuality gene, an alcoholism gene or a “homicide” gene all cause the related behavior and thus the individual has no control over it. People are just victims of the gene lottery. In actuality, genetics, environment and our will determine the course of our lives. Genes do not act in isolation.

In this view, people are seen not as persons but as products or living machines. This type of thinking increasingly permeates the secular media and results in a flight from personal responsibility and yields no hope or need for change. Biological determinism also leads to prejudice toward those with “bad genes” and further fuels the eugenic movement.

Commercialization

A patent is a “grant made by a government that confers upon the creator of an invention the sole right to make, use, and sell that invention for a set period of time.”¹⁴ Any creation short of a “full human being” qualifies for a patent. Traditionally it was thought that the US Constitution states that patenting, which can be made on any creation short of a “full human being”, is essential to research and innovation. The Constitution protects full human beings from being patented due to the 13th amendment prohibiting slavery but this is beginning to change. Recently a Midwestern university was given a patent on all the products of a cloning process which could include human cloning.

There were already 7,800 applications for patents on genetic life forms by 1990.

Distributive Justice

Who will benefit from genetic advances? Genetic diagnoses and therapies are expensive. A genetically engineered growth hormone cost \$30,000 a year in 1993. It costs a family \$80,000 to have their child gain one inch in height.

Vaccination and public health measures will save many more lives at less cost. The investment in human growth hormone in 1993 totaled \$300 million.

Is it fair to spend so much on technologies that will affect only a few when the same investment would save more lives on a worldwide basis by finding a vaccine for an epidemic killer like malaria or gastroenteritis?

What You Can Do: Know What the Bible Says



Privacy & Discrimination

- **Every being of human origin is a person.**

"...For in the image of God has God made man." —Genesis 9:6

- **Biblical justice says there is value and dignity in weaker human beings.**

"On the contrary, those parts of the body that seem to be weaker are indispensable, and the parts that we think are less honorable we treat with special honor. And the parts that are unpresentable are treated with special modesty, while our presentable parts need no special treatment. But God has combined the members of the body and has given greater honor to the parts that lacked it, so that there should be no division in the body, but that its parts should have equal concern for each other. If one part suffers, every part suffers with it; if one part is honored, every part rejoices with it. Now you are the body of Christ, and each one of you is a part of it." —1 Corinthians 12:22-27

- **God tells us to love everyone the same.**

"You have heard that it was said, 'Love your neighbor and hate your enemy.' But I tell you: Love your enemies and pray for those who persecute you." —Matt 5:43-44

Stigmatization

We must understand that, no matter what we think of ourselves, we have great worth to God. Because He loves us we should have love for ourselves and recognize that God has a plan for our lives in spite of any genetic predisposition or disease.

- **Every individual is important to God.**

"For God so loved the world that he gave his one and only Son, that whoever believes in him shall not perish but have eternal life." —John 3:16

- **We can have confidence in God's goodness in every situation.**

"I am still confident of this: I will see the goodness of the Lord in the land of the living." —Psalm 27:13

Eugenics

- **Children have special value to God. He especially cares for the unborn.**

"Did you not pour me out like milk and curdle me like cheese, clothe me with skin and flesh and knit me together with bones and sinews? You gave me life and showed me kindness, and in your providence watched over my spirit." —Job 10:11-12

"Did not he who made me in the womb make them? Did not the same one form us both within our mothers?" —Job 31:15

"For you created my inmost being; you knit me together in my mother's womb. I praise you because I am fearfully and wonderfully made; your works are wonderful, I know that full well. My frame was not hidden from you when I was made in the secret place. When I was woven together in the depths of the earth." —Psalm 139:13-15

"Listen to me, O house of Jacob, all you who remain of the house of Israel, you whom I have upheld since you were conceived, and have carried since your birth. Even to your old age and gray hairs I am he, I am he who will sustain you. I have made you and I will carry you; I will sustain you and I will rescue you." —Isaiah 46:3-4

- **God despises those that destroy the weak.**

"... Those whose teeth are swords and whose jaws are set with knives to devour the poor from the earth, the needy from among mankind." —Proverbs 30:14

- **Child sacrifice is condemned.**

"Do not give any of your children to be sacrificed to Molech, for you must not profane the name of your God. I am the Lord." —Leviticus 18:21

- **We are not allowed to shed innocent blood.**

"If you really change your ways and your actions and deal with each other justly, if you do not oppress the alien, the fatherless or the widow and do not shed innocent blood in this place, and if you do not follow other gods to your own harm, then I will let you live in this place, in the land I gave your forefathers for ever and ever." —Jeremiah 7:5-7



"In you men accept bribes to shed blood; you take usury and excessive interest and make unjust gain from your neighbors by extortion. And you have forgotten me, declares the Sovereign Lord. I will surely strike my hands together at the unjust gain you have made and at the blood you have shed in your midst." —Ezekiel 22: 12-13

- **We are to be good stewards of the resources and knowledge given to us by God.**

For with wisdom comes much sorrow; the more knowledge, the more grief. —Ecclesiastes 1:18

Then I turned my thoughts to consider wisdom, and madness and folly...I saw that wisdom is better than folly, just as light is better than darkness. —Ecclesiastes 2:12,13

Biological Determinism

- **People have free will and have control over their own passions.**

"The acts of the sinful nature are obvious: sexual immorality, impurity and debauchery; idolatry and witchcraft; hatred, discord, jealousy, fits of rage, selfish ambition, dissensions, factions and envy; drunkenness, orgies, and the like. I warn you, as I did before, that those who live like this will not inherit the kingdom of God. But the fruit of the Spirit is love, joy, peace, patience, kindness, goodness, faithfulness, gentleness and self-control. Against such things there is no law." —Galatians 5:19-23

- **Through God's spirit we can overcome temptation.**

"No temptation has seized you except what is common to man. And God is faithful; he will not let you be tempted beyond what you can bear. But when you are tempted, he will also provide a way out so that you can stand up under it." —1 Corinthians 10:13

Commercialization & Distributive Justice

- **We should give to others.**

"Our desire is not that others might be relieved while you are hard pressed, but that there might be equality. At the present time your plenty will supply what they need, so that in turn their plenty will supply what you need. Then there will be equality, as it is written: 'He who gathered much did not have too much, and he who gathered little did not have too little.'" —2 Corinthians 8:13-15

- **We should treat others as our neighbors.**

"But he wanted to justify himself, so he asked Jesus, 'And who is my neighbor?' In reply Jesus said: 'A man was going down from Jerusalem to Jericho, when he fell into the hands of robbers. They stripped him of his clothes, beat him and went away, leaving him half dead. A priest happened to be going down the same road, and when he saw the man, he passed by on the other side. So too, a Levite, when he came to the place and saw him, passed by on the other side. But a Samaritan, as he traveled, came where the man was; and when he saw him, he took pity on him. He went to him and bandaged his wounds, pouring on oil and wine. Then he put the man on his own donkey, took him to an inn and took care of him. The next day he took out two silver coins and gave them to the innkeeper. 'Look after him,' he said, 'and when I return, I will reimburse you for any extra expense you may have.' Which of these three do you think was a neighbor to the man who fell into the hands of robbers?' The expert in the law replied, 'The one who had mercy on him.'" Jesus told him, 'Go and do likewise.'" —Luke 10:29-37

- **Everyone deserves just treatment because they are made in God's image.**

"For God so loved the world that he gave his one and only Son, that whoever believes in him shall not perish but have eternal life." - John 3:16

"Do to others as you would have them do to you." -Luke 6:31

4. Resources

Christian Legal Society

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Life Issues Institute

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Cincinnati, OH 45239
513.729.3600
513.729.3636
www.lifeissues.org

Concerned Women for America

1015 Fifteenth St. NW Suite 1100
Washington, DC 20005
202.488.7000
www.cwfa.org

Focus on the Family

8605 Explorer Drive
Colorado Springs, CO 80902
719.531.3328
800.A-FAMILY
www.family.org

Family Research Council

801 G. Street NW
Washington, DC 20001
202.393.2100
800.225.4008
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Endnotes

- ¹ Human Genome Project Information. November 25, 2002. <http://www.ornl.gov/hgmis/project/about.html>
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- ⁶ *Am. J. Public Health* 68:1116-1120, (1978).
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