

Handout

Is Molecular Genetics Redefining What it Means to be Human? – Richard Buggs

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A) Humans as a distinct category, separate from the rest of the animal kingdom

1) We humans like to think of ourselves as special, set apart from the rest of the animal kingdom by our ability to talk, write, build complex structures, and make moral distinctions. But when it comes to genes, humans are so similar to the two species of chimpanzee that physiologist Jared Diamond has called us "the third chimpanzee." A quarter-century of genetic studies has consistently found that for any given region of the genome, humans and chimpanzees share at least 98.5% of their DNA. This means that a very small portion of human DNA is responsible for the traits that make us human, and that a handful of genes somehow confer everything from an upright gait to the ability to recite poetry and compose music.

Gibbons (1998) *Science* 281:1432 – 1434 Which of Our Genes Make Us Human?

2) Humans differ from both common chimps and pygmy chimps in about 1.6 per cent of their (our) DNA, and share 98.4 per cent. Gorillas differ somewhat more, by about 2.3 per cent, from us and from both of the chimps.

Let us pause to let some of the implications of these momentous numbers sink in.

... the chimpanzees' closest relative is not the gorilla but the human. Traditional taxonomy has reinforced our anthropocentric tendencies by claiming to see a fundamental dichotomy between mighty man, standing alone on high, and the lowly apes all together in the abyss of bestiality. Now future taxonomists may see things from the chimpanzees' perspective: a weak dichotomy between slightly higher apes (the *three* chimpanzees, including the 'human chimpanzee') and slightly lower apes (gorilla, orang-utan, gibbon). The traditional distinction between 'apes' (defined as chimps, gorillas, etc.) and humans misrepresents the facts.

Diamond (1993) *The Third Chimpanzee* In Cavalieri and Singer (eds.), *The Great Ape Project* (New York: St. Martin's Griffin), pp. 89-101.

3) The accumulating DNA evidence provides an objective nonanthropocentric view of the place of humans in evolution. We humans appear as only slightly remodeled chimpanzee-like apes.

Wildman et al. (2003) *Proc Natl Acad Sci U S A.* 100: 7181–7188.

4) ...from the point of view of hemoglobin structure, it appears that gorilla is just an abnormal human, or man an abnormal gorilla, and the two species form actually one continuous population.

Zuckerandl (1963) "Perspectives in Molecular Anthropology" pp243-272 in *Classification and Human Evolution*. Washburn (Ed.). Aldine, Chicago IL.

5) From any point of view other than that properly specified, that is of course nonsense. What the comparison seems really to indicate is that in this case, at least, hemoglobin is a bad choice and has nothing to tell us about affinities, or indeed tells us a lie.

G. G. Simpson (1964) *Science* 146:1535-1538

6) ...all the biochemical methods agree in showing that the genetic distance between humans and the chimpanzee is probably too small to account for their substantial organismal differences...The molecular similarity between chimpanzees and humans is extraordinary because they differ far more than sibling species in anatomy and way of life. Although humans and chimpanzees are rather similar in the structure of the thorax and arms, they differ substantially not only in brain size but also in the anatomy of the pelvis, foot, and jaws, as well as in relative lengths of limbs and digits. Humans and chimpanzees also differ

significantly in many other anatomical respects, to the extent that nearly every bone in the body of a chimpanzee is readily distinguishable in shape or size from its human counterpart. Associated with these anatomical differences there are, of course, major differences in posture, mode of locomotion, methods of procuring food, and means of communication. Because of these major differences in anatomy and way of life, biologists place the two species not just in separate genera but in separate families. So it appears that molecular and organismal methods of evaluating the chimpanzee-human difference yield quite different conclusions.

King and Wilson (1975) *Science* 188:107-116

7) Rights - and responsibilities - are exclusively human attributes. DNA is beside the point. To concede so much to biology risks taking such privileges away from ourselves...Chimps may resemble *Homo sapiens* in a tedious and literal sense, but in everything that makes us what we are *H sapiens* is unique indeed. Biology, in its proof of our physical similarity to other primates, underlines its own irrelevance. The double helix does not diminish us but places mankind on a metaphysical, moral, and ethical peak of its own.

Prof Steve Jones (UCL) *Daily Telegraph* 26/06/2007

8) For many, many years, the 1% difference served us well because it was underappreciated how similar we were. Now it's totally clear that it's more a hindrance for understanding than a help.

Pascal Gagneux, zoologist at UC San Diego, quoted in Cohen (2007) *Science* 316: 1836

9) Our results imply that humans and chimpanzees differ by at least 6% (1,418 of 22,000 genes) in their complement of genes, which stands in stark contrast to the oft-cited 1.5% difference between orthologous nucleotide sequences.

Demuth et al (2006) *PLoS ONE* 1:e85

10) ...humans and chimpanzees are known to differ in major chromosomal rearrangements involving 10 of 22 human autosomal chromosomes.

Rieseberg and Livingstone (2003) *Science* 300:267-268.

11) Although the majority of regions in our genome are most closely related to chimpanzees and bonobos, a non-trivial fraction is more closely related to gorillas

Paabo (2003) *Nature* 421:409-412.

12) Could researchers combine all of what's known and come up with a precise percentage difference between humans and chimpanzees? "I don't think there's any way to calculate a number," says geneticist Svante Pääbo, a chimp consortium member based at the Max Planck Institute for Evolutionary Anthropology in Leipzig, Germany. "In the end, it's a political and social and cultural thing about how we see our differences."

Cohen (2007) *Science* 316:1836

13) Sameness/otherness is a philosophical paradox that is resolved by argument, not by data. Genetic data tells us precisely what we already knew: that humans are both very similar to and different from the great apes.

Marks (2002) *What it Means to be 98% Chimpanzee*, University of California Press pp22-23

B) Humans as responsible moral agents

14) Marriage Woes? Husband's Genes May Be At Fault

Men who are lacking in the romance department may have a new excuse to offer their wives or significant others: They can blame it on their DNA.

A new study in the Proceedings of the National Academy of Sciences suggests that a certain gene variant found in some men is linked to less bonding and more trouble in a long-term relationship.

Greenfieldboyce (2008). NPR. All Things Considered, September 2, 2008

15) Time and time again, scientists have claimed that particular genes or chromosomal regions are associated with behavioral traits, only to withdraw their findings when they were not replicated. "Unfortunately," says Yale's [Dr. Joel] Gelernter, "it's hard to come up with many findings linking specific genes to complex human behaviors that have been replicated."

...The 1960s to 80s saw a succession of claims about genetic variants associated with aggression, manic depression, schizophrenia, and psychosis. All were announced with great fanfare; all were greeted unskeptically in the popular press; all are now in disrepute.

... the interaction of genes and environment is much more complicated than the simple "violence genes" and intelligence genes" touted in the popular press. The same data that show the effects of genes, also point to the enormous influence of nongenetic factors."

Mann (1994) Genes and behavior. *Science* 264:1687

16) Men... may have peculiar corruptions, which either by their natural constitution or education and other prejudices, have got deep rooting in them. This also is to be found out by him, who would not enter into temptation. Unless he know it, unless his eyes be always on it, unless he observe its motions, advantages, it will continually be entangling him.

John Owen (1658) *Of Temptation*

C) Human minds having a non-physical component

17) ...one major hope is that the differences between the [human and chimpanzee] sequences will reveal the genetic basis for our mental and linguistic capacities... "It will tell us what makes us human," claims Yoshiyuki Sakaki of the University of Tokyo's Institute of Medical Science

Cyranoski (2002) *Nature News* feature 418:910-912

18) We share with chimpanzees some—but not all—mental functions, some of which are shared with other species as well. As the publication of the chimpanzee genome reveals, we also share a good deal of our DNA. Unfortunately, we are virtually in the dark when it comes to understanding how genes build minds....we are woefully ignorant about how genes build brains, and how the electrical activity of the brain builds thoughts and emotions.

Hauser (2005) *Nature* 437:60-63

19) Gene expression in the human brain has attracted particular interest because many of the phenotypic differences that set humans apart from other organisms have a cognitive component. One might therefore expect the human brain to have diverged more from the brains of other animals than other organs have diverged from their homologues. Contrary to this expectation, however, gene expression in the brain has diverged less than other organs analysed to date.

Khaitovich et al. (2006) *Nature Reviews Genetics* 7, 693-702

20) It is the awareness of right and wrong, along with the development of language, awareness of self, and the ability to imagine the future, to which scientists commonly refer when trying to enumerate the special qualities of *Homo sapiens*... Consider a major example of the force we feel from the Moral Law – the altruistic impulse, the voice of conscience calling us to help others even if nothing is received in return... Agape, or selfless altruism...is quite frankly a scandal to reductionist reasoning. It cannot be accounted for by the drive of selfish genes to perpetuate themselves. Quite the contrary: it may lead humans to make sacrifices that lead to great personal suffering, injury, or death, without any evidence of benefit. And yet, if we carefully examine that inner voice we sometimes call conscience, the motivation to practice this kind of love exists within all of us, despite our frequent efforts to ignore it... If the law of human nature cannot be explained away as cultural artifact or evolutionary by-product, then how can we account for its presence? There is truly something unusual going on here. To quote Lewis "If there was a controlling power outside the universe, the only way in which we could expect it to show itself would be inside ourselves as an influence or a command trying to get us to behave in a certain way. And that is just what we do find in ourselves. Surely this ought to arouse our suspicions."

Francis Collins (2006) *The Language of God*. Free Press. New York. pp23-29

21) The physiologist studies the development of the first cell of each new human baby into a full-grown adult. The evolutionary biologist studies the forces which have formed the genetic structure of such a first cell. But relatively seldom do either of these scientists point out that their descriptions and explanations cover only the evolution of the physical characteristics of man, and that they give no account of the evolution of the most important characteristics of man—the characteristics of his conscious life, his feelings and desires, hopes and beliefs, those characteristics in virtue of his possession of which we treat men, and think that we ought to treat men, as totally different from machines...While I shall attempt to describe the differences between men and machines, I shall not be able fully to explain their origin; and indeed I shall produce an argument to show that it is most unlikely that anyone will ever be able fully to explain their origin in terms of a normal scientific explanation. The prospects for a fully explanatory super-science which embraces the mental world as well as the physical world are poor. But we must not fall into the trap of believing that that which we cannot explain (at any rate by a normal scientific pattern of explanation) does not exist. The conscious life evidently exists—that we have sensations and thoughts, feelings and hopes is the most evident thing that there is.

Swinburne (1997) *The Evolution of the Soul* Oxford University Press, p.3

22) All 'reduction' of one science to another dealing with apparently very disparate properties has been achieved by [the] device of denying that the apparent properties (i.e. the 'secondary qualities' of colour, heat, sound, taste, etc.) with which one science dealt belonged to the physical world at all. It siphoned them off to the world of the mental. But then, when you come to face the problem of the sensations themselves, you cannot do this. If you are to explain the sensations themselves, you cannot distinguish between them and their underlying causes and only explain the latter. In fact, the enormous success of science in producing an integrated physico-chemistry has been achieved at the expense of separating off from the physical world colours, smells and tastes, and regarding them as purely private sensory phenomena. The very success of science in achieving its vast integrations in physics and chemistry is the very thing which has made apparently impossible any final success in integrating the world of the mind and the world of physics.

Swinburne (1997) *The Evolution of the Soul* Oxford University Press, p.191

23) Explaining "humanness" is a vague and broadly philosophical question, not easily approached using the genome alone.

Varki and Altheide (2005) *Genome Research* 15:1746-1758

Table 1: Differences between the human and chimpanzee genome

Type of difference	Difference	Cumulative difference
Non-alignable ¹	~24%	24%
Single Nucleotide Polymorphism ¹	~3% (of 76%)	27.9%
Indertion/deletion ¹	1.23% (of 76%)	29.5%
Copy Number Variation (>20kb) ²	2.7%	—
Gene Complement ³	6%	—

¹Nature 437:69-87 ²Nature 437:88-93 ³PLoS ONE 1:e85

Table 2: Some phenotypic traits of humans for comparison with those of great apes (from Table 1, Varki and Altheide (2005) *Genome Research* 15:1746-1758)

- **LIFE HISTORY**
 - Secondary Altriciality
 - Helplessness of the Newborn
 - Prolonged Helplessness of Young
 - Extended Care of Young
 - Childhood
 - Adolescence
 - Age at First Reproduction
 - Longevity
- **REPRODUCTIVITY BIOLOGY**
 - Concealed Ovulation
 - Virgin Breast Development
 - Female Pituitary Menopause
 - Placentophagy
 - Female Labia Majora
 - Vaginal Hymen
 - Baculum (Penis Bone)
 - Sperm Count
 - Copulatory Plug
- **EMBRYOLOGY**
 - Early Fetal Wastage/Aneuploidy
 - Hydatiform Molar Pregnancy
 - Umbilical Cord Length
- **PREGNANCY/PARTURITION**
 - Cephalo-pelvic Disproportion
 - Duration of Labor
 - Maternal Mortality in Childbirth
 - Pain During Childbirth
 - Need for Assistance with Childbirth
 - Neonatal Cephalhematoma
- **POSTNATAL DEVELOPMENT**
 - Late Closure of Cranial Sutures
 - Duration of Infant Arousal
 - Inconsolable Infant Crying
 - Infant-Caregiver Attunement
 - Maternal-Infant Eye-To-Eye Gaze
- **ANATOMY**
 - Sagittal Crest of Skull
 - Brow Ridge
 - Protuberantia Menti (Chin)
 - Length of Sphenoid Sinus
 - Choroid Plexus Biondi Bodies
 - Inner Ear Canal Orientation
 - Apical Phalangeal Tufts
 - Age of Pelvic Bone Fusion
 - Bone Cortex Thickness
- Laryngeal Position
- Pharyngeal Air Sacs
- Ear Lobes
- Sexual Body Size Dimorphism
- Lacrimal Gland Structure
- Visible Whites of the Eyes
- Small/Large Intestine Length Ratio
- Meningeal Artery Source
- **BIOMECHANICS**
 - Bipedal Gait
 - Adductive Thumb
 - Skeletal Muscle Strength
 - Hand-Eye Coordination
 - Fine Motor Coordination
- **ORGAN PHYSIOLOGY**
 - Aldosterone Response to Posture
 - Salt-Wasting Kidneys
 - Ability For Sustained Running
 - Voluntary Control of Breathing
 - Ability to Dive Underwater
 - Diving Reflex
 - Ability to Float/Swim
 - Emotion Lacrimation
 - Salt Content of Tears
 - Olfactory Sense
- **BIOCHEMISTRY**
 - Placental Alkaline Phosphatase
 - N-Glycolylneuraminic Acid Expression
 - Alpha 2-6-Linked Sialic Acid Expression
- **ENDOCRINOLOGY**
 - Thyroid Hormone Metabolism
- **PHARMACOLOGY**
 - Methylation of Inorganic Arsenic
- **ANATOMIC PATHOLOGY**
 - Cortical Neurofibrillary Tangles
- **CLINICAL PATHOLOGY**
 - Erythrocyte Sedimentation Rate
 - Serum Alkaline Phosphatase Level
 - RBC and Serum Folate
 - Serum Vitamin B12/B12 Binding
 - Total Leukocyte Count
 - Absolute Neutrophil Count
 - Absolute Lymphocyte Count
- **DENTAL BIOLOGY/DISEASE**
 - Canine Tooth Diastema
 - Canine Tooth Dymorphism
 - Tooth Enamel Thickness
 - Retromolar Gap
 - Third Molar Impaction
 - Dental Eruption Sequence/Timing
- **MEDICAL/SURGICAL DISEASES**
 - HIV Progression to AIDS
 - *P. falciparum* malaria
 - Viral Hepatitis B/C Complications
 - Influenza A Infection Severity
 - Incidence of Carcinomas
 - Hemorrhoids
 - Varicose Veins
 - Pelvic Phleboliths
 - Foamy Virus (Spumavirus) Infections
 - Sexually Transmitted Diseases
- **IMMUNOLOGY**
 - Sialoadhesin on Macrophages
- **SKIN BIOLOGY AND DISEASE**
 - Eyebrows
 - Eccrine Sweat Glands
 - Acne Vulgaris
 - Subcutaneous Fat
 - Body Lice
- **NUTRITION**
 - Frugivory
 - Carnivory
 - Aquatic Foods
 - Underground Foods
 - Cooking
- **NEUROANATOMY**
 - Relative Brain Size
 - Direct Cortical Projections
 - Relative Volume of Frontal Cortex
 - Relative Volume of Corpus Callosum
 - Relative Volume of Cerebellum
 - % of Brain Growth Complete at Birth
 - Rate of Postnatal Brain Growth
- **NEUROBIOLOGY**
 - Population Distribution of Handedness
 - Postnatal Dendritic Growth

- Postnatal Synapse Formation
- Cortical Synapse Density
- Cortical Neuron Density
- Dendrites Per Neuron
- Synapses Per Neuron
- Adult Neurogenesis
- Cingulate Cortical Spindle Neurons
- Finger Tip Sensory Nerve Endings
- **NEUROCHEMISTRY**
- Brain Aromatisation of Testosterone
- Tyrosine Hydroxylase Heterogeneity
- **MENTAL DISEASE**
- Schizophrenia
- Bipolar Psychosis
- Autism
- Suicide
- **BEHAVIOR**
- Control of Facial Expressions
- Planning Ahead
- Intentional Deception
- Deliberately Delaying Gratification
- Long-Range Transport of Materials
- Secondary Tool-Making
- Mechanical Multi-Tasking
- Physical Abuse of the Young
- Torture
- Organized Warfare
- Adult Play
- Symbolic Play
- Abuse of Other Animals
- Inter-Group Coalition Formation

- Use of Containers
- Care of Infirm and Elderly
- Grandparenting
- Home Base
- Control of Fire
- Food Preparation
- Organized Gathering of Food
- Domestication of Animals
- Domestication of Plants
- Altruistic Punishment
- Peace-Making
- Somnambulism
- Mind-Altering Drug Use
- **COGNITIVE CAPACITY**
- Declarative Memory
- Imitative Learning
- Teaching
- Symbolic Representation
- Awareness of Death
- Awareness of the Past
- Awareness of the Future
- Theory of Mind
- Theory of Other Minds
- Empathy
- Numeracy
- **COMMUNICATION**
- "Parentese" Sounds
- Infant "Protoconversations"
- Gestural Communication
- Symbolic Communication
- Semantics
- Grammar and Syntax
- Recursion
- Writing
- **SOCIAL ORGANIZATION**
- Institutions
- Social Conventions
- Governments
- Enforcement Through

- Sanctions
- **CULTURE**
- Composition of Art
- Composition of Music
- Composition of Rhythms
- Death Rituals
- Clothing (Covering of Body Parts)
- Rites of Passage
- Genocide
- Competitive Sports
- Practicing of Skills
- Physical Modifications of the Body
- Inheritance of Resources and Status
- Rhythmic Dance
- Sculpture
- Belief in Supernatural/Religion
- Body Adornment
- Childbirth Customs
- Sexual Intercourse in Private
- Gift-Giving
- Hospitality
- Intertwining (e.g. weaving)
- Meal Times
- Poetry
- Property
- Construction of Shelters
- Taboos
- Taxonomy of Species
- Trade
- Measurement of Time
- Weapons
- Toys