NOTES

**Genomics** is an interdisciplinary field of biology focusing on the structure, function, evolution, mapping, and editing of genomes.

SLIDE:

**(Human) Genome**: the complete set of genes or genetic material that makes us.

-Our biology has become another form of information technology.

SLIDE:

Researchers are comparing the DNA of 1.1 million people (looking for patterns of variations).

-they Map the genome and look for variations (that cause a given trait).

SLIDE: **HEIGHT**

Let’s say you have a bunch of tall people

-and you want to find out what makes them tall, genetically

-you take the genomes of all of the tall people

-and you feed the billions and billions (6.4) of letters (base pairs, As, Ts, C, Gs) into an ML model. (the whole genome sequence of an individual)

-ML algorithms will train on the data, scanning and looking for patterns, like “what do these tall people have in common?”

-a model might learn that: a bunch of them have a certain mutation at location 398, 078,

- and a bunch of them also have a mutation at location 1m 212

- and another mutation at location 63,812

An ML model learned that there are about 20,000 locations in the genome that influence height

Once you have this trained model, you can then feed in a new individual’s genome (lust the genome into the learned model)

-it evaluates the 20,000 locations (for example) to make a prediction of height within about an inch

-it can certainly predict whether someone will be tall, average or

-researchers don’t know what those genetic variations are doing, but they just know that the model indicated that there is something different happening in these places for the taller people than for the other people.

-it can also predict other heritable things like diabetes and breast cancer

-some traits like eye color or diseases like cystic fibrosis, are relatively straight forward to identify because they are expressed in single gene mutations.

-but, for the most part, the human genome functions in very complex ways that we don’t fully understand.

-but AI (applied to genomics) is making this more accessible

- algorithms are run to analyze genome-wide patterns in order to identify specific genes and further our understanding of the human genome.

-with more data, our understanding will improve

-it is predicted that 2 billion human genomes will be sequenced over the next 10 years.

-as the technology has become a lot less expensive

-the cost of sequencing a full human genome has fallen from $100 million in 2001 to around just $700 today.

**SLIDE**: IVF

- a controversial application of all of this is with IVF

SLIDE: **Automating embryo selection**

The system was trained using 12,000 photos of embryos, all photographed exactly 110 hours after fertilization. Each of the photos was marked with a grade by a trained embryologist, to identify the good embryos from the bad. Moreover, the actual outcome resulting from these embryos was known at the time of the research, so that was an additional feature trained on. The resulting algorithm has shown a 97% consistency with what humans would decide would be a viable embryo.

-IVF makes up around 1.5% of all births in the United States

-increasingly, genetic testing is done where the genetic code of the cell is screened

-genetic testing can ensure that offspring are healthy before being implanted

-the most common genetic testing involves checking that the number of chromosomes is normal (screened against Down’s Syndrome)

-but it also has been used for screening an embryo for its gender

-selecting an offspring’s gender accounts for 9% of pre(implantation) genetic testing in the U.S., although it’s illegal in other countries like the U.K., Austria, Chile and China.

-beyond gender, embryos are being selected for increasingly complex traits

--they can say that embryo #4 will have diabetes, or #2 has a strange outlier for (some trait)

-further, research is being done to screen for intelligence and personality

-What’s the genetic architecture of human intelligence?

-but since complex traits like IQ are made from hundreds of genes, embryo selection for these complex traits will elicit a probability, rather than a binary option. For example, you’ll be able to select that your child will have a 70% probability/likelihood of being intelligent, etc.

-As IVF and pre genetic testing grow to circumvent more and more genetic mutations, prospective parents will increasingly opt to give their children the best chance at a healthy life. For example, if you could screen out Type 1 diabetes or cancer, would you?

-If the doctor said that the risks are a lot higher (for this trait) than for all of your other embryos, what would you do

-couples may be faced with choosing which embryos are the best to use

Embryo selection/screening enables you to select future offspring based on the genetic makeup of the embryo.

**SLIDE**: Gene editing

-but, to take it a step further, in the future, rather than just choosing an embryo, you’ll be able to manipulate your embryo’s genetic code.

-advances in gene editing will lead us into genetic manipulation far beyond healthcare

-you may be able to choose your skin color or superior hearing or vision, and of course intelligence

-the big problem is that all of the genetic data collected is on white people of European descent

-wouldn’t apply to non-whites

-more data is being collected

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**SLIDE: Squirrel AI**

-uses AI to break a given subject into knowledge points. Their relationships—how they build on each other and overlap—are encoded in a “knowledge graph,”

**SLIDE: Curriculum Knowledge Graph**

Interdisciplinary network of Biology

A student begins a course of study with a short diagnostic test to assess how well she understands key concepts. If she correctly answers an early question, the system will assume she knows related concepts and skip ahead. Within 10 questions, the system has a rough sketch of what she needs to work on, and uses it to build a curriculum. The system can directly identify the gaps in understanding

As she studies, the system updates its model of her understanding and adjusts the curriculum accordingly. As more students use the system, it spots previously unrealized connections between concepts. The machine-learning algorithms then update the relationships in the knowledge graph to take these new connections into account.

-this is called adaptive learning

-it has been very successful in China in lifting test scores (especially math)

-it is deeply personalized teaching.

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NLP – sentiment analysis

The restaurant made an error on her order. It's a good thing it wasn't my child who nearly choked because that restaurant would be out of business. I will never go there again!

The error that the restaurant made wasn’t so great. It’s a good thing it wasn’t my child. I would have choked with laughter from everything on the menu and that restaurant would be out of business.

Tsao,

“Cracking the code of Facial Recognition”

Cal Tech