NOTES

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

**(Human) Genome**: the complete set of genes or genetic material present in a cell or organism.

-nearly 3 million letters if our genetic code

What’s the genetic architecture of human intelligence?

Find/Map the genome and looking for variations (that cause that trait).

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

Genome-wide association studies.

**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)

-an 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)

-The model can evaluate, what’s at this location, and this location and this location,

-it evaluates the 20,000 locations (for example) and research has shown that it can make a prediction of height within about an inch (it’s not perfect because other things influence height beyond the genes), but it can certainly predict whether someone will be tall, average or short

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

-it can predict other inheritable things like diabetes and breast cancer

**INTELLIGENCE**

-Interestingly, in the last year or so, researchers are now looking at these genome-wide associations as a way to predict intelligence

-Intelligence (cognitive ability) is a complex trait with a lot of genes involved

But also, environment, diet, and other variables matters also

-research suggests that ones genes are 20-50% responsible for intelligence

-the hope is that if you had enough data, a trained model could learn how to crudely predict cognitive performance from your genotype alone

-few individuals from the 1.1 million people in the study had IQ data available

-however, most did have educational attainment (the number of years that an individual had been in school) (e.g. HS diploma, PhD)

-it turns out that the genetic pattern that predicts educational attainment was pretty good at predicting IQ

-similar correlations (.3-.4): SAT to GPA in college, educational attainment to IQ

**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.

Now, this research is being applied (a company called Genomic Prediction) to genetic (polygenic) testing of embryos, particularly relating to IVF (every year, 1 million babies are born world-wide through IVF)

-several embryos are produced

-one or two cells are removed from an embryo through a biopsy

-the genetic code of the cell is screened

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

-sequences a part of the genome of an embryo

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

-they’re looking at predicting intelligence

-but right now, the only thing that is reported is if the embryo is an outlier at risk for intellectual disability (embryo #4 has an unusually large number of the variance that depress intelligence)

-But intelligence is coming

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

-How many of you would screen an embryo for a certain trait if it were possible?

Would you rather go with a gut-feeling based on the shape or on a genetic evaluation?

(The risks are a lot higher (for this trait) than for all of your other embryos)

-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**

Once the knowledge points are set, they are paired with video lectures, notes, worked examples, and practice problems. Their relationships—how they build on each other and overlap—are encoded in a “knowledge graph,” also based on the master teachers’ experience.

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. 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.

-the ultimate goal is deeply personalized teaching.

Squirrel has offered some validation of its system. In October 2017, for example, a self-funded four-day study with 78 middle school students found that the system was better on average at lifting math test scores than experienced teachers teaching a dozen or so kids in a traditional classroom.

“… the instruction is very targeted; the system can directly identify the gaps in my understanding.”

Squirrel is doing adaptive learning, which is about “understanding exactly what students know and don’t know.”

**SLIDE: Curriculum Knowledge Graph**

**Interdisciplinary network of Biology**

The next Figure 2 shows the interdisciplinary network of Biology (in green) departments and research groups. Several units are identified and their relationships with other groups of others disciplines. For instance, the Ecology and Environment cluster is linked with the Earth Science websites (purple), the Cytology and Physiology groups are linked with the Medicine research groups (white) and the Biochemistry and Molecular Biology set is connected with the Chemistry sites (light red). However, the Genetics group does not make relationship with other disciplines.

-the ultimate goal is deeply personalized teaching.

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**SLIDE**: AI Autopilots

-the average flight of a Boeing plane involves only seven minutes of human-steered flight, which is typically reserved only for takeoff and landing.

**SLIDE**: DNA Hacking

-Our biology is becoming another form of information technology.

-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.

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

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

-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 patterns: (e.g., height?)

-the hope is to identify specific genes and further our understanding of the human genome.

**SLIDE**: IVF

-these advances are leading to embryo screening/selection for increasingly complex traits

-doctors can sequence an embryo’s genome before implantation, to do genetic testing,

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

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

-selecting embryos for preferable traits is already becoming acceptable. Selecting an offspring’s gender accounts for 9% of preimplantation genetic testing in the U.S., although it’s illegal in other countries like the U.K. and China.

-as our understanding grows, we will be able to screen for diseases such as cancer

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

-but since hundreds of genes make up complex traits like IQ, 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 tall, intelligent, etc.

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

-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?

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

**SLIDE**: Gene editing

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

-gene editing is more precise and affordable than ever

-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.

NLP – sentiment analysis

The restaurant made a great 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.