Descifrando el genoma humano: un tesoro o la caja de Pandora?

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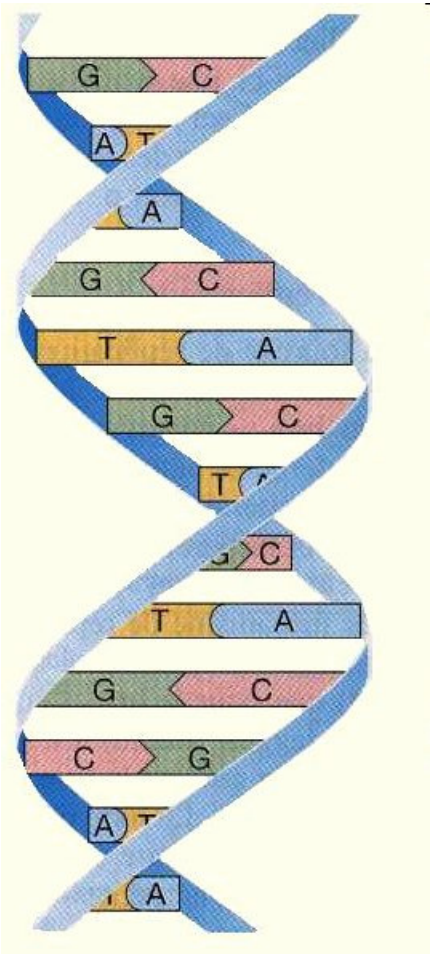
Instituto de Biología y Medicina Experimental

Proyecto Genoma Humano



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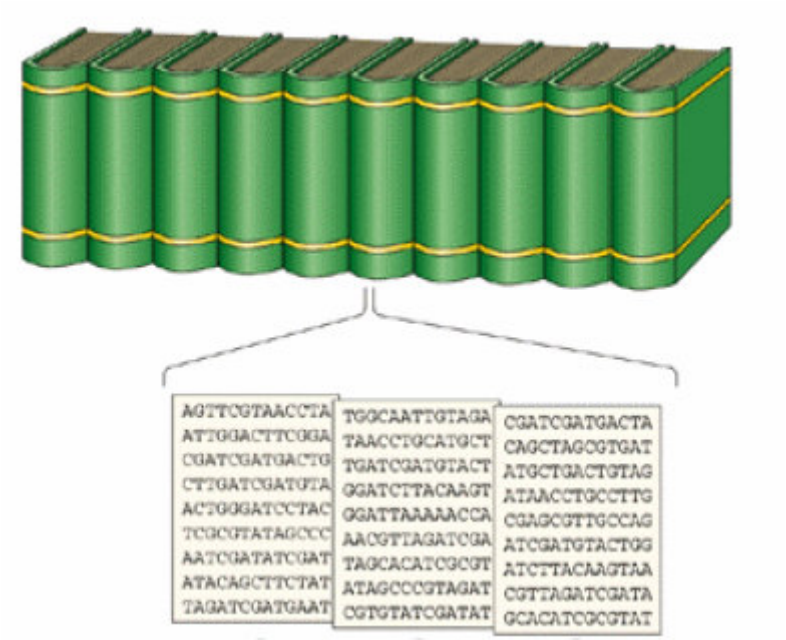
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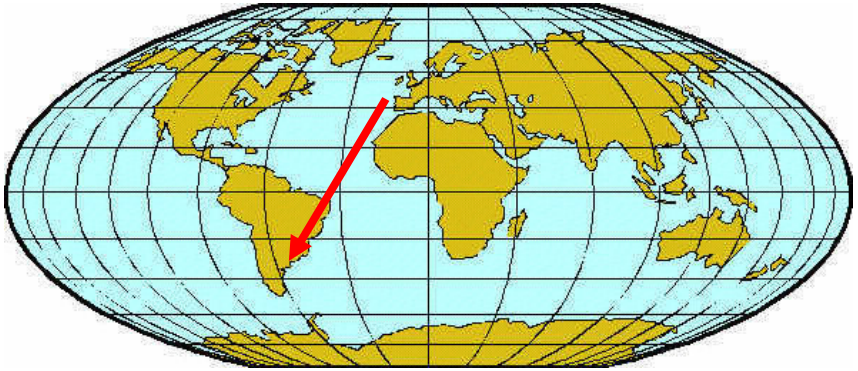
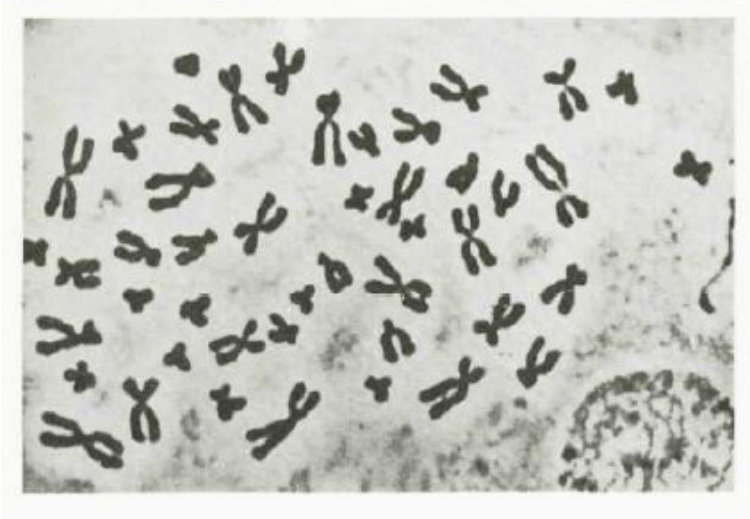
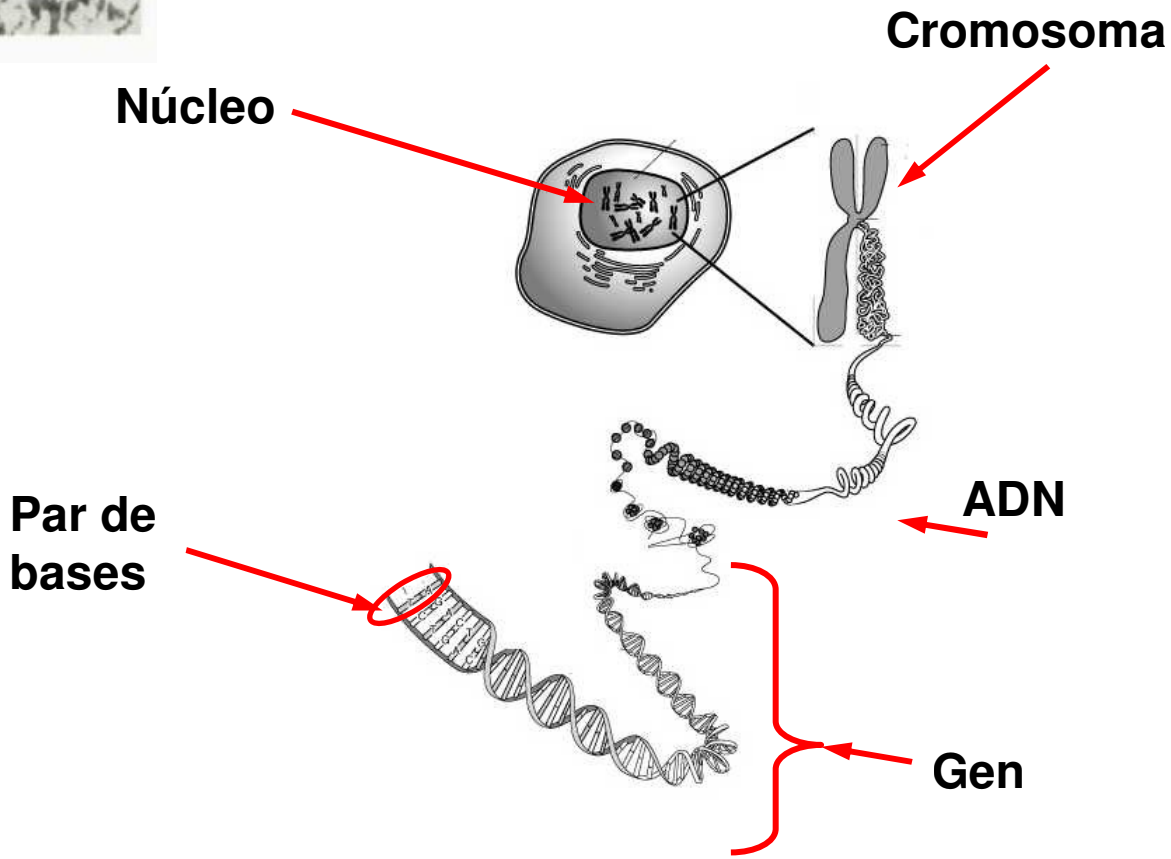





Cromosoma
Núcleo
ADN
Par de bases
Gen

Almacenamiento en una biblioteca y el Genoma

Unidad de información	Libro	Gen
Tamaño	Miles de libros	30000 genes
Lenguaje	Alfabeto 26 letras	Alfabeto 4 letras
Unidades	Palabras de varias letras	Codones de 3 letras. Hay 20 palabras
Organización	Estantes por tema	Cromosomas, información al azar
Búsqueda	Catálogos	No conocidos
Circulación información	Se sacan los libros	Se copian los genes

Los tesoros del Genoma

- ***1) La similitud del genoma de las distintas especies es increíble***

- ***2) El número de genes no permite explicar la complejidad de los organismos vivos***

Los tesoros del Genoma

- ***3) ADN basura o ADN que no codifica.***
- ***4) Tenemos pocos genes pero muchas proteínas.***
- ***5) Tenemos 200 genes cuyos parientes más cercanos están en las bacterias.***

Los tesoros del Genoma

- ***6) Hay más de 99% de similitud. Lo que nos distingue entre humanos son los polimorfismos.***

- ***7) Toda la vida sobre la tierra es una sola***

Usos destacados o posibles del genoma

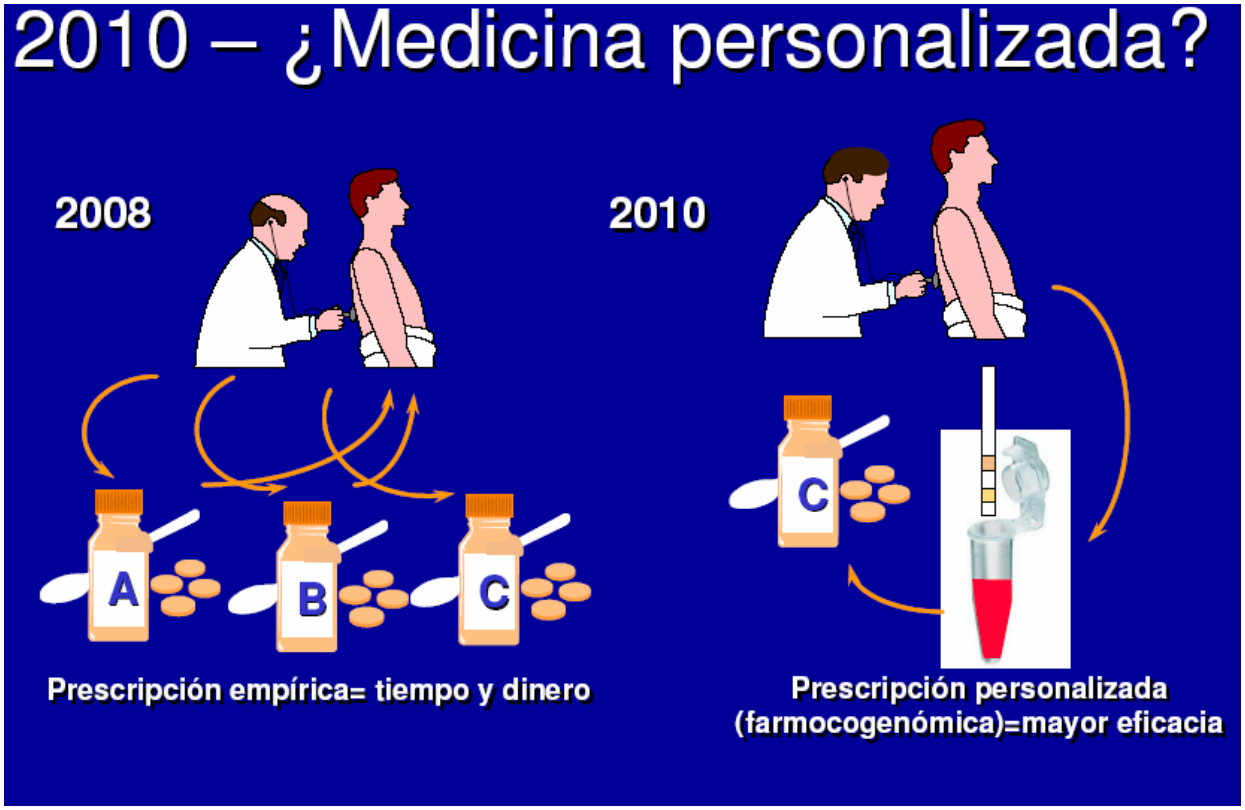
¿Para qué nos serviría este gran libro de cuatro letras que se repiten?

- ***Predecir la enfermedad***
- ***Diagnosticar la enfermedad***
- ***Intervenir en la enfermedad***

Tabla VII. Tasas de detección de mutaciones en diferentes patologías

enfermedad		*incidencia*	*gen*	*tasa de detección de mutación*
monogénica	fibrosis quística	1:4000	*CFTR*	98 %
	distrofia Duchenne muscular	1:4000	*DMD*	90 %
	síndrome X-frágil	1:4000	*FMR*	100 %
	huntington	1:5000	*HD*	100 %
	hemofilia A	1:10000	*F8C*	90 %
	fenilcetonuria	1:10000	*PAH*	99 %
	riñón poliquístico	1:1500	*PKD1* *PKD2*	15 %
cáncer hereditario	ovario-mama	1:4000	*BRCA1*	60 %
			BRCA2	35 %
	síndrome Li-Fraumeni		*p53*	50 %
	ataxia-teleangiectasia		*ATM*	70 %
	poliposis de colon familiar	1:4000	*APC*	87 %
	no-poliposis de clon hereditaria	1:2000	*MLH1*	33 %
			MSH2	12 %
trastornos cardiovasculares	hipercolesterolemia familiar	1:500	*LDLR*	60 %
	hiperlipidemia		*APOE*	10 %

Predecir infectividad y respuesta terapéutica.
Antropología molecular
Farmacogenética Farmacogenómica



Lo Epigenético

- ***Las huellas digitales***
- ***El sistema inmune***
- ***Susceptibilidad a la enfermedad***
- ***El sistema nervioso central***

La caja de Pandora

- garantizar la privacidad de los datos genéticos,
- el respeto a la diversidad genética,
- y la correcta utilización del diagnóstico genético.

Del gen al medicamento. La prospectiva islandesa

Mapping the Icelandic Genome

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http://www.decode.com/

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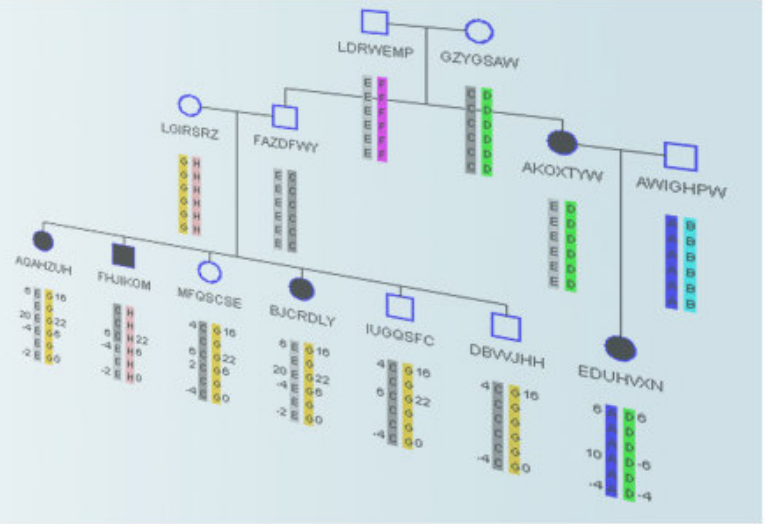

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Listo

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NATURE|Vol 464|1 April 2010

The balance scale

A matter of life and death.

Shelly Li

Imagine a balance scale with all your sins on one side and your morals on the other.

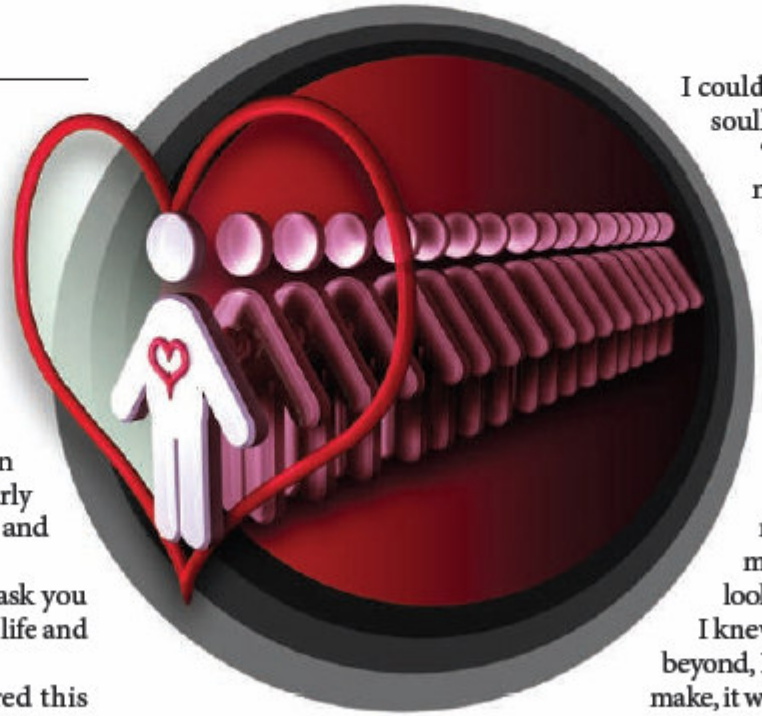
I'm not a Godly man, nor do I attend church or pray for my sins to be dumped like yesterday's garbage. But if I had to imagine my balance scale, I'd say that it's pretty even.

That is, until five years ago: when the doctor diagnosed me with early sepsis and multiple organ failure, and challenged my morals for a cure.

I guess the question I want to ask you is: where do your morals go when life and death hang in the balance?

Let me tell you how I answered this question.

After my fall from the roof in 2045, I spent close to a week in a hospital bed.



JACEY

"We will do all that we can to keep the toxins at bay," he said. "In the best scenario, I could live a long, healthy life … it felt soulless.

"I will give you BioClone's number, and the name of my friend," Dr Pei finally said. "Tell him that I sent you, and he will help you from there." The doctor smiled sadly as he handed me a slip of paper. "The choice, however, is yours."

As soon as he finished speaking, the door slid open again, and Annie's face appeared before me. Her puffy eyes were red, her mascara smeared. But the way she looked at me, that hopeless stare that I knew would haunt me to death and beyond, I realized that this choice I had to make, it would not be for me alone. Could I compromise my morals to save Annie from the pain that my death would cause?

This is my story, and my existence is my

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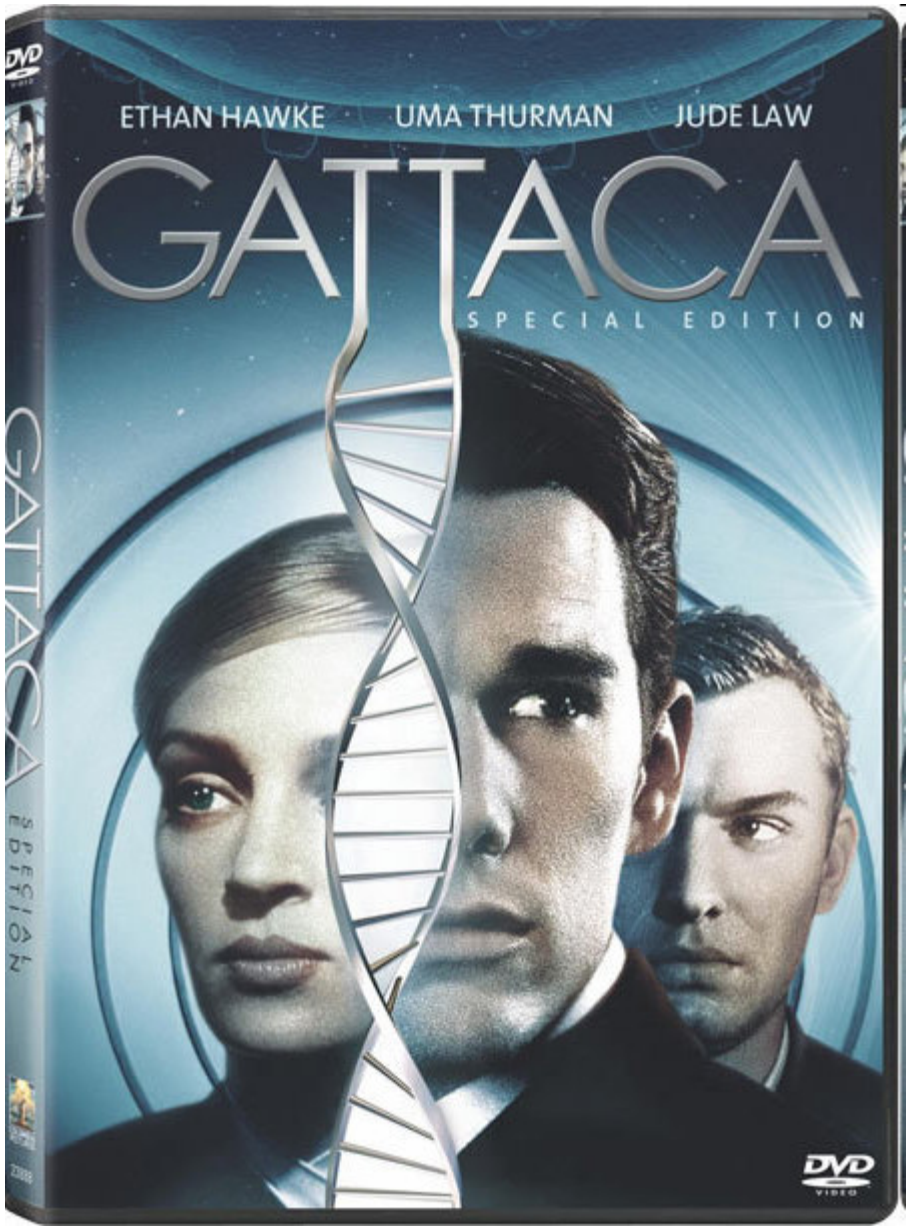
ETHAN HAWKE
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JUDE LAW
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SPECIAL EDITION
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No hay gen para el espíritu humano

No hay un gen del destino.

"Créeme, en tu corazón brilla la estrella de tu destino."

Johann Christoph Friedrich von Schiller