




SOSTITUIRÀ

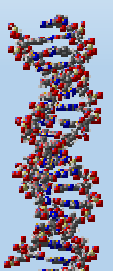
?????

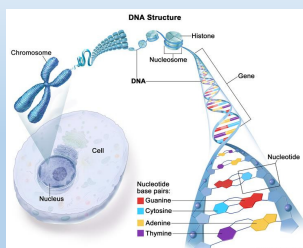
Maria Francesca Armentano, PhD

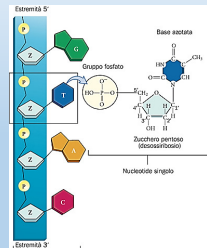
Potenza - Matera
15-16 Aprile 2019











Il DNA è un polinucleotide, cioè un polimero di nucleotidi, ognuno formato da tre parti:

- uno zucchero C₅ detto **desossiribosio**
- un gruppo **fosfato**
- una base **azotata**

Esistono quattro diversi tipi di basi azotate.

L'**adenina (A)** e la **guanina (G)** sono caratterizzate da un doppio anello e sono chiamate *purine*. La **timina (T)** e la **citosina (C)** sono caratterizzate da un anello singolo e sono chiamate *pirimidine*.

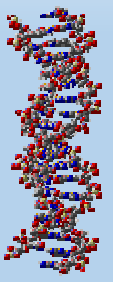
Il DNA e l'RNA, gli acidi nucleici, sono polimeri di nucleotidi aventi requisiti adatti per funzionare come materiale genetico

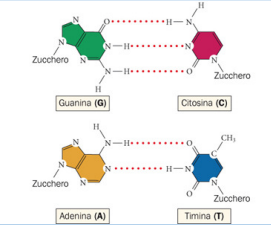




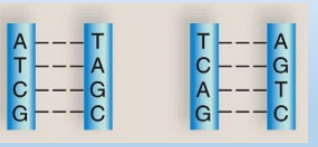


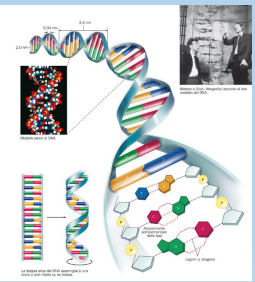




La molecola del DNA ha la forma di una **doppia elica** di nucleotidi in cui la base **A** è sempre in coppia con la base **T**, e la base **C** con la base **G**.





L'appaiamento complementare delle basi azotate suggerisce che il DNA è una **molecola a doppio filamento**, simile a una scala a pioli in cui i montanti sono costituiti dallo **scheletro zucchero-fosfato**, e i pioli dalle **basi accoppiate unite da legami idrogeno**.

La molecola del DNA ha la forma di una doppia elica









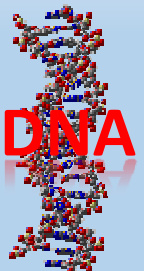

ORGANIZZAZIONE CROMOSOMICA DEI GENI

ORGANIZZAZIONE STRUTTURALE DEI GENI EUCARIOTICI

TRASCRIZIONE

SINTESI PROTEICA

REPLICAZIONE DNA



CONTROLLO TRASCRIZIONALE DELL'ESPRESSIONE GENICA

RIPARAZIONE E RICOMBINAZIONE DEL DNA

CONTROLLO POST-TRASCRIZIONALE DELL'ESPRESSIONE GENICA







DNA INSPIRED ART







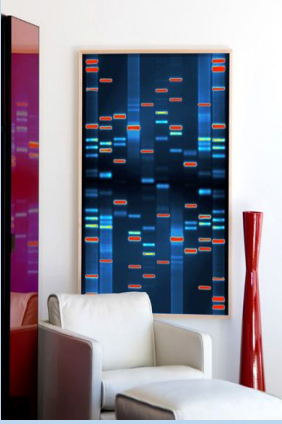








DNA INSPIRED ART

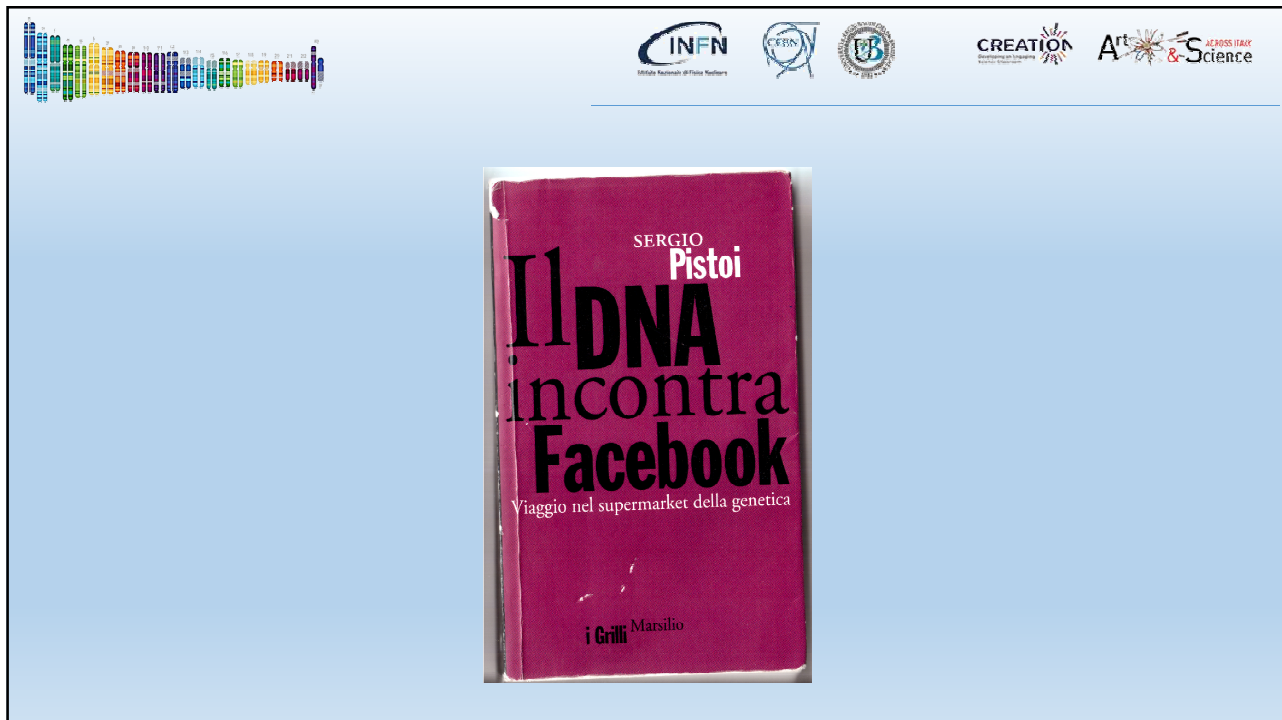












METTIAMO IN FILA LE LETTERINE A, C, T, G.....
.....IL METODO SANGER DI SEQUENZIAMENTO DEL DNA

Nature, vol. 265, February 24 1977

articles

Nucleotide sequence of bacteriophage ϕ X174 DNA

F. Sanger, G. M. Air*, B. G. Barrell, N. L. Brown*, A. R. Coulson, J. C. Fiddes, C. A. Hutchinson III*, P. M. Slocumbe* & M. Smith*

MRC Laboratory of Molecular Biology, Hills Road, Cambridge CB2 2QH, UK

Proc. Natl. Acad. Sci. USA
Vol. 74, No. 12, pp. 5463-5467, December 1977
Biochemistry

DNA sequencing with chain-terminating inhibitors
(DNA polymerase/nucleotide sequences/bacteriophage ϕ X174)

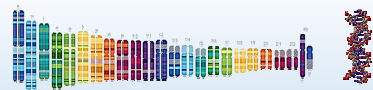
F. SANGER, S. NICKLEN, AND A. R. COULSON
Medical Research Council Laboratory of Molecular Biology, Cambridge CB2 2QH, England

120 130
GAT AAAT CT GGTCTT ATTTCC

Frederick Sanger
1958 1980

**Il metodo di Sanger ha
segnato l'inizio della
ricerca genomica**

[http://www.kaltura.com/index.php/extwidget/preview/partner_id/362071/uiconf_id/8603321/entry_id/1_10wlcjtl/embed/auto?&flashvars\[streamerType\]=auto](http://www.kaltura.com/index.php/extwidget/preview/partner_id/362071/uiconf_id/8603321/entry_id/1_10wlcjtl/embed/auto?&flashvars[streamerType]=auto)



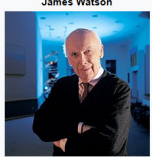









Human Genome Project Information Archive 1990-2003



James Watson



Francis Collins

- Consortio pubblico creato per portare a termine la decifrazione del DNA umano
- Una delle imprese scientifiche più importanti della storia umana: determinazione della sequenza di circa tre miliardi di lettere, che si succedono in sequenze ininterrotte di milioni, formando frasi, capitoli, volumi scritti in CODICE GENETICO
- Mille ricercatori di sei paesi (USA, Regno Unito, Germania Francia Giappone, Cina)
- Concorrenza della Celera di Craig Venter
- Costo: **TRE MILIARDI DI DOLLARI!!!!**



GenBank Overview
GenBank® is the first genetic sequence database, an annotated collection of all publicly available DNA sequences. (Bioinformatics Research, 2012 Jan 4;10(1):105-123). GenBank is part of the International Nucleotide Sequence Database Collaboration, which comprises the DNA DataBank of Japan (DDBJ), the European Nucleotide Archive (ENA), and GenBank at NCBI. These three organizations exchange data on a daily basis.

DIGITALIZZAZIONE DEL PATRIMONIO GENETICO

In realtà si era solo all'inizio, poiché tali sequenze di lettere andavano **STUDIATE, ANALIZZATE e, SOPRATTUTTO, SFRUTTATE!!!!**

https://web.ornl.gov/sci/techresources/Human_Genome/index.shtml

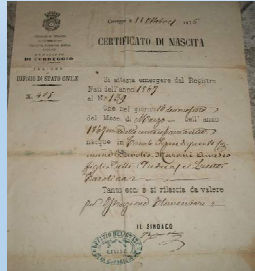









GENOMICA: studio dell'intero DNA dell'individuo






GENOMICA DI MASSA

Carta di identità biologica: lista dei punti del DNA che ci rendono unici e diversi dagli altri, e la ricerca di queste differenze individuali è la ragione che spiega la **nascita e l'esplosione** della **GENOMICA PERSONALIZZATA**, UNA SCIENZA CHE SI RIVOLGE IN PRIMIS A **PERSONE SANE**, al vasto pubblico di **CONSUMATORI**, e non ad una specifica categoria



TIME Magazine
Invention of the Year 2008

23andMe®

TIME
Best Inventions of 2008

From a genetic testing service to an unrivaled public data system to the world's first missing information — here are TIME's picks for the top inventions of 2008.

INVENTION OF THE YEAR
1. The Retail DNA Test
By Anita Hendon — Wednesday, Oct. 29, 2008

Before meeting with Anne Wojcicki, co-founder of a consumer gene testing service called 23andMe, I knew just three things about her: she's pregnant, she's married to Google's Sergey Brin, and she went to Yale. But after an hour chatting with her in the small office she shares with co-founder Linda Avey at 23andMe's headquarters in Mountain View, Calif., I know some things no Internet search could reveal: coffee makes her giddy, she has a fondness for organic shoes and Irish-limed bread, and her unborn son has a 90% chance of inheriting a high risk for Parkinson's disease.

Learning and sharing your genetic secrets are at the heart of 23andMe's commercialized new service — a \$99 saliva test that estimates your predisposition for more than 2,600 traits and conditions ranging from baldness to blindness. Although

http://content.time.com/time/specials/packages/article/0,38804,1852747_1_854493,00.html

<https://www.23andme.com/en-int/>



Ancestry Service

Welcome to you

Experience your ancestry in a new way! Get a breakdown of your global ancestry by percentages, connect with DNA relatives and more. [learn more](#)

\$99

[add to cart](#)

Health + Ancestry Service

Welcome to you

Get an even more comprehensive understanding of your genetics. Receive 125+ online reports on your ancestry, traits and health - and more. [learn more](#)

\$199

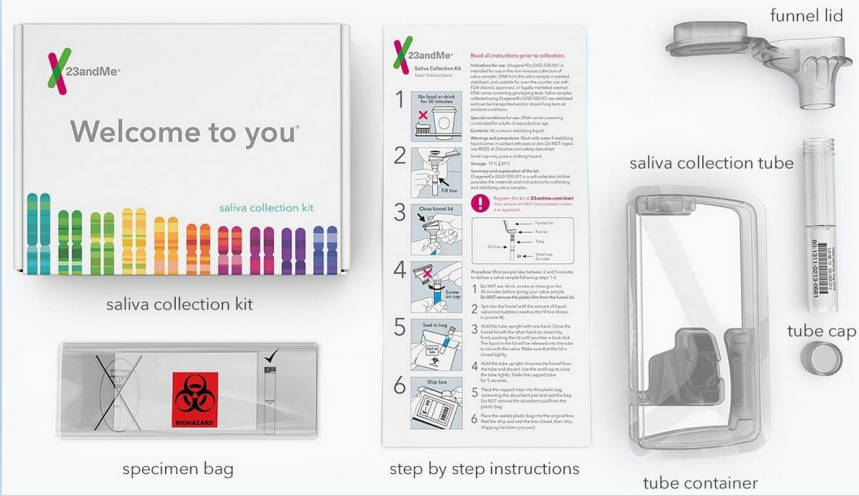
[add to cart](#)

Human Genome Project
2003 - 13 anni
3 miliardi di dollari

Craig Venter
2007 - 2 anni
100 milioni di dollari

James Watson
2008 - 4 mesi
2 milioni di dollari

<https://www.23andme.com/en-int/>



saliva collection kit

specimen bag

step by step instructions

funnel lid

saliva collection tube

tube cap

tube container

- Estrazione DNA dalle cellule della mucosa buccale
- Alcuni enzimi lo 'digeriscono', ovvero lo tagliuzzano in frammenti di poche migliaia di lettere
- Sequenziamento (Sanger) o Microarray (Chip a DNA)



Dal sequenziamento e dall'analisi delle varianti (SNP, single nucleotide polymorphism):

- Suscettibilità genetica ad una serie di malattie / risposta individuale ad alcuni farmaci
- Analisi di tratti individuali, anche connessi al comportamento (altezza, colore degli occhi, attitudini sportive, memoria, reazioni a cibi, a odori, riservato o affabile ecc ecc.)

Health Predisposition reports*

10+ reports

Type 2 Diabetes (Powered by 23andMe Research)

Genetic likelihood for a disorder of blood sugar regulation

1,000+ variants in many genes; variants found in many ethnicities

BRCA1/BRCA2 (Selected Variants)

Genetic risk based on a limited set of variants for breast, ovarian and other cancers

3 variants in the BRCA1 and BRCA2 genes; relevant for Ashkenazi Jewish descent

Age-Related Macular Degeneration

Genetic risk for a form of adult-onset vision loss

2 variants in the ARMS2 and CFH genes; relevant for European descent

Celiac Disease

Genetic risk for gluten-related autoimmune disorder

2 variants near the HLA-DQB1 and HLA-DQA1 genes; relevant for European descent

Familial Hypercholesterolemia

Genetic risk for very high cholesterol, which can increase the risk for heart disease

24 variants in the LDLR and APOB genes; relevant for European, Lebanese, Old Order Amish descent

G6PD Deficiency

Genetic risk for a form of anemia

1 variant in the G6PD gene; relevant for African descent

Late-Onset Alzheimer's Disease

Genetic risk for a form of dementia

1 variant in the APOE gene; variant found and studied in many ethnicities

Parkinson's Disease

Genetic risk for a form of movement impairment

2 variants in the LRRK2 and GBA genes; relevant for European, Ashkenazi Jewish, North African Berber descent

Traits reports

30+ traits

Ability to Match Musical Pitch

Asparagus Odor Detection

Back Hair (available for men only)

Bald Spot (available for men only)

Bitter Taste

Cheek Dimples

Cilantro Taste Aversion

Cleft Chin

Dandruff

Earlobe Type

Early Hair Loss (available for men only)

Earwax Type

Eye Color

Fear of Heights

Finger Length Ratio

Freckles

Hair Photobleaching (hair lightening from the sun)

Hair Texture

Hair Thickness

Light or Dark Hair

Ecc ecc.....

ASSUMI UN RUOLO PIÙ ATTIVO NELLA TUA SALUTE

PREPARATI AD AFFRONTARE MALATTIE SERIE

I TUOI GENI SONO LA STRADA PER UN BENESSERE MIGLIORE



«Bugiardino»

Genetic Health Risk reports tell you about genetic variants associated with increased risk for certain health conditions. They do not diagnose cancer or any other health conditions or determine medical action.



Having a risk variant does not mean you will definitely develop a health condition. Similarly, you could still develop the condition even if you don't have a variant detected. It is possible to have other genetic risk variants not included in these reports.



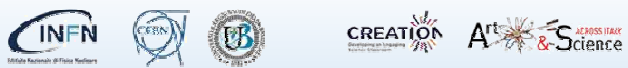
Factors like lifestyle and environment can also affect whether a person develops most health conditions. Our reports cannot tell you about your overall risk for these conditions, and they cannot determine if you will or will not develop a condition.



These reports do not replace visits to a healthcare professional. Consult with a healthcare professional for help interpreting and using genetic results. Results should not be used to make medical decisions.

↓

NO CONTROLLI
(Food and Drug Administration, FDA
European Society Human Genetics, ESGH)

«Bugiardino»






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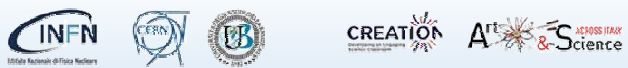
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Il dubbio è:

Veramente voglio conoscere se sono PREDISPOSTO ad una patologia, vivendo, forse inutilmente, nell'ansia????



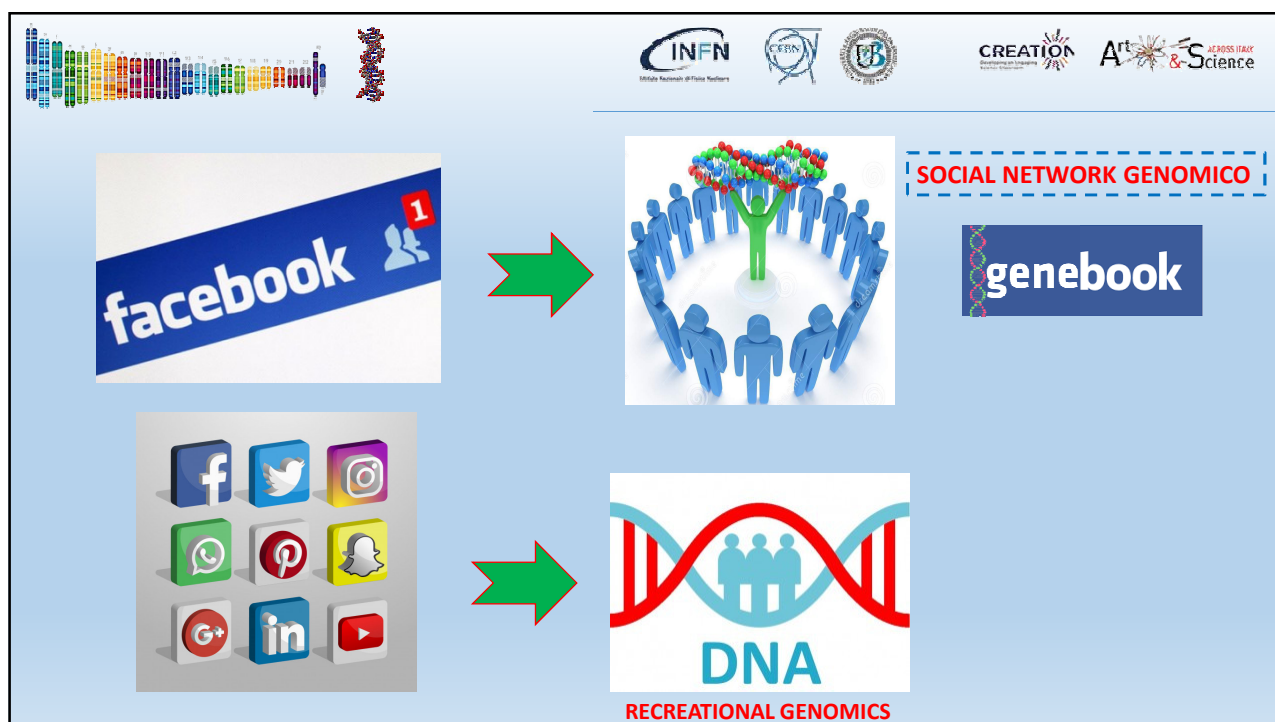


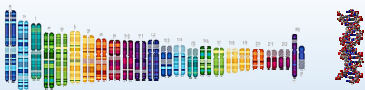
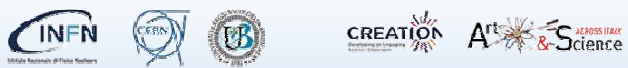


DNA Relative Finder

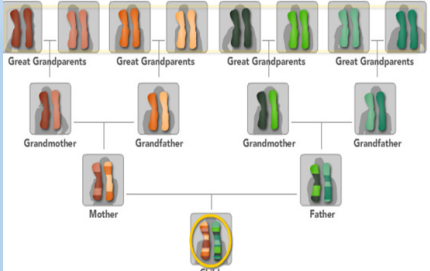
- Discover DNA relatives from around the world.
- Choose to message and meet relatives - maybe even hear new family stories, share photos and get a better understanding of your family's history.

..... IL TROVAPARENTI!!!!!!

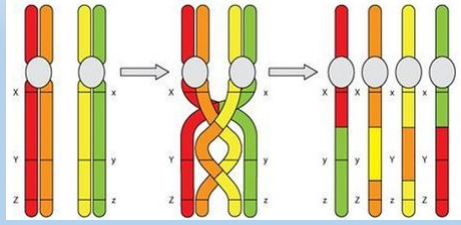


➤ **Profilo genealogico**



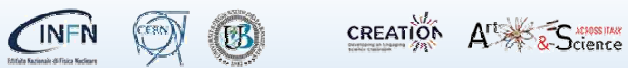
Great Grandparents
Grandmother
Grandfather
Mother
Father
Child



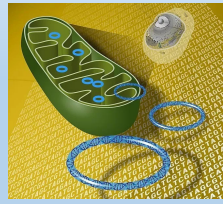
CROSSING OVER

IL DNA SI CONSERVA NEL TEMPO, PER GENERAZIONI, COME FOSSE UNA SCATOLA NERA INSERITA NEI NOSTRI CROMOSOMI!!!!!!
GENEALOGIA MOLECOLARE

DI GENERAZIONE IN GENERAZIONE, CROMOSOMI SEMPRE PIÙ «DILUITI»
 ↓
 SI RISALE AL MASSIMO ALLA 10° GENERAZIONE (~ 200 ANNI)

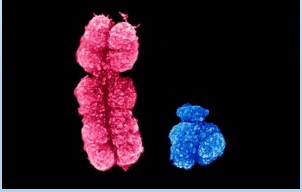
...MA PER LA STORIA DELL'UOMO 200 ANNI SONO UN BATTITO DI CIGLIA!!!!!!



DNA MITOCONDRIALE



CROMOSOMA Y

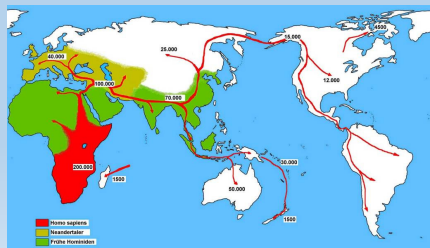


POSSIBILE RIPERCORRERE LA NOSTRA STORIA NEL CORSO DI MILLENNI

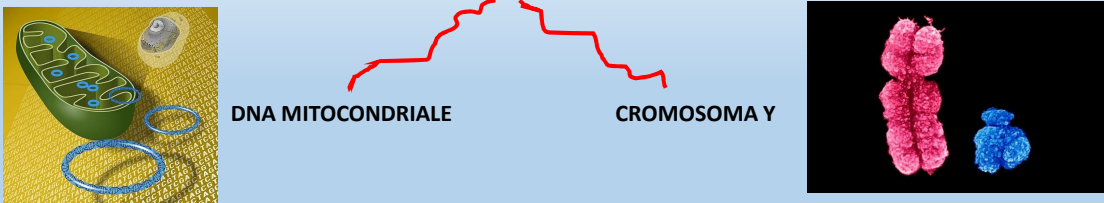
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GENOGRAFIA

Homo sapiens: il lungo viaggio dall'Africa



...MA PER LA STORIA DELL'UOMO 200 ANNI SONO UN BATTITO DI CIGLIA!!!!!!



DNA MITOCONDRIALE

CROMOSOMA Y

**TUTTI GLI UOMINI NON AFRICANI CHE OGGI
POPOLANO LA TERRA
DISCENDONO DA QUEI SAPIENS FUGGITIVI**



Ancestry \$99

35+ reports

Ancestry Composition
Ancestry Detail Reports (33 reports)
Maternal Haplogroup
Paternal Haplogroup
Neanderthal Ancestry
Your DNA Family

Jamie King

European	47.4%
• Iberian	19.7%
• Ashkenazi Jewish	0.5%
• Scandinavian	0.2%
• Broadly Southern European	21.1%
• Broadly Northwestern European	0.3%
• Broadly European	5.5%
East Asian & Native American	41.4%
• Native American	34.4%
• Han	< 0.1%
• Manchurian & Mongolian	< 0.1%
• Southeast Asian	< 0.1%
• Broadly East Asian	0.5%
• Broadly East Asian & Native American	6.8%
Sub-Saharan African	5.2%
• West African	4.5%
• East African	< 0.1%
• African Hunter-Gatherer	< 0.1%
• Broadly Sub-Saharan African	0.6%
Western Asian & North African	1.2%
• North African & Arab	1.0%
• Broadly Western Asian & North African	0.2%
Unassigned	0.4%

See all 150+ populations

Your DNA comes from around the world.

23andMe Find your roots

47.1% Northwest European
23.2% Chinese
21.2% Southeast Asian
2.6% Southern European

Alcune riflessioni:

genebook

IN BASE AL NOSTRO DNA

Offerta di prodotti mirati

Possibilità di discriminazioni

Brevettato, in California, un sistema di
«Pubblicità mirata, basata sul genoma».....
.....Ingenuity Systems!!!!!!

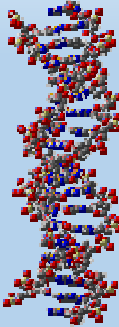
CONCETTO DI PRIVACY DIVENTERÀ ASSOLUTAMENTE CRUCIALE!!!!

Quale sarà l'evoluzione della genomica di massa/social network genomico?

Top Non-Game Apps by Worldwide Downloads for 2018

Overall Downloads	App Store Downloads	Google Play Downloads
1 WhatsApp	1 TikTok	1 WhatsApp
2 Messenger	2 YouTube	2 Messenger
3 Facebook	3 WhatsApp	3 Facebook
4 TikTok	4 Instagram	4 TikTok
5 Instagram	5 Messenger	5 Instagram
6 UC Browser	6 Facebook	6 SHAREit
7 SHAREit	7 Google Maps	7 UC Browser
8 YouTube	8 Netflix	8 Vigo Video
9 Snapchat	9 Gmail	9 Snapchat
10 Vigo Video	10 Snapchat	10 YouTube

...anche le **applicazioni genomiche** entreranno a far parte della nostra vita di tutti i giorni



Grazie per l'attenzione!!!!