Genteknologi.
Hvor er vi nå - og om ti år?

Dag Undlien

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Oslo Universitetssykehus

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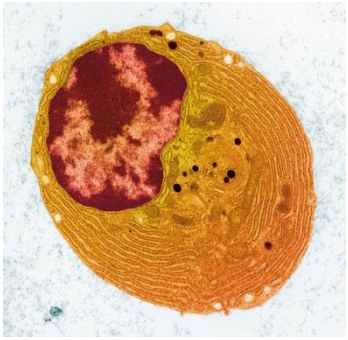
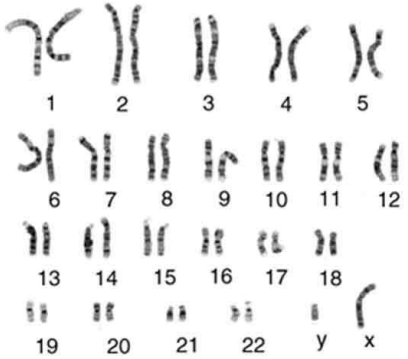
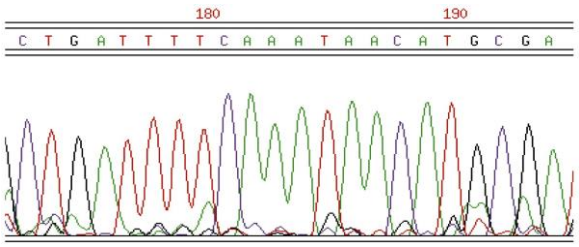
Genomet - oppskriftsboken







- 3 milliarder tegn (basepar)
- 4 bokstavers alfabet C G A T

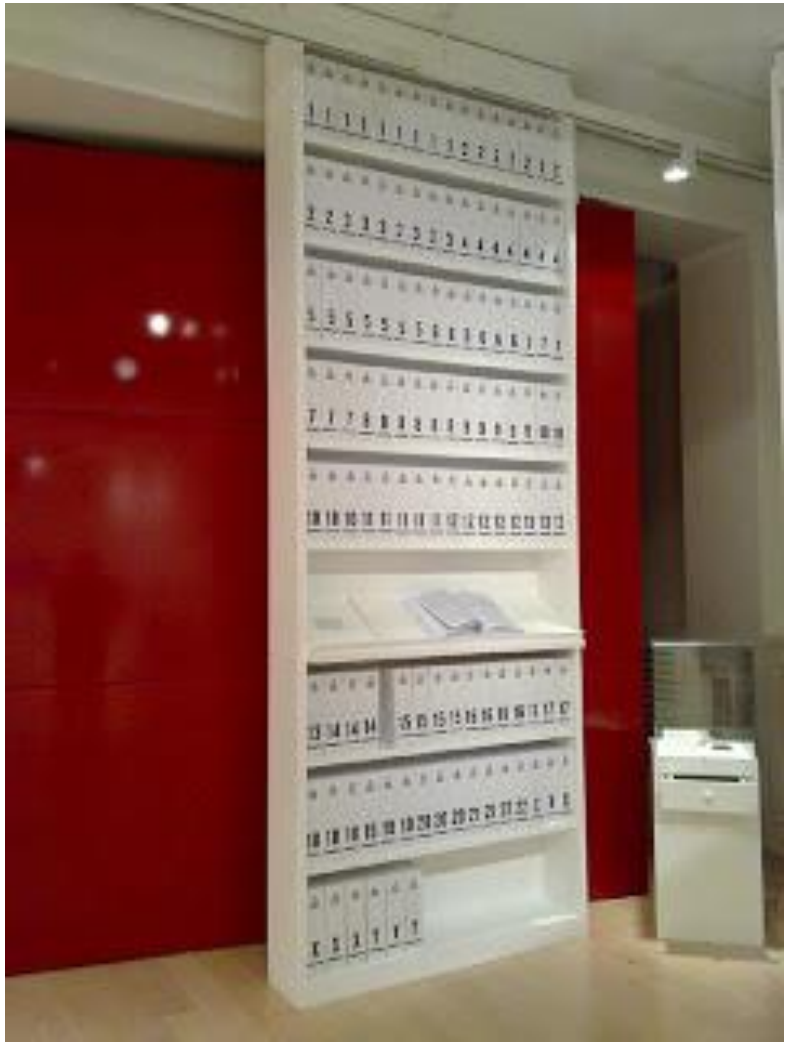
- 23 kapitler = kromosomer

- 20.000 oppskrifter = gener

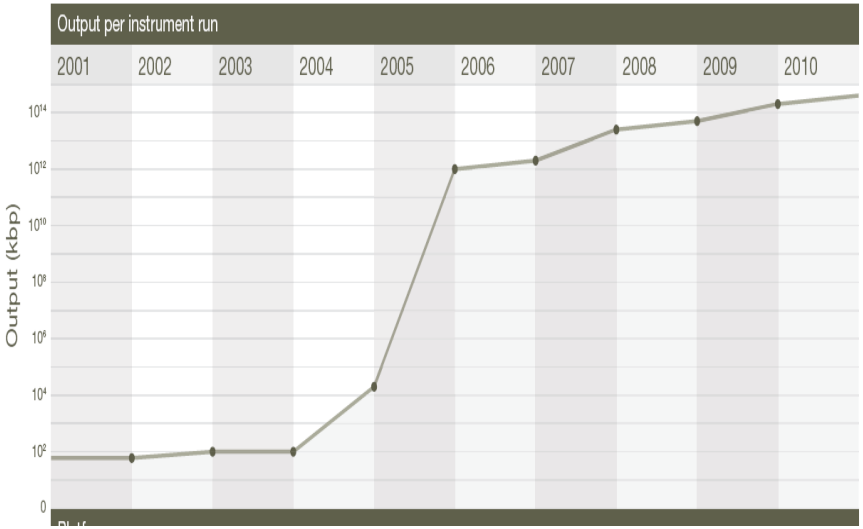
- DNA sekvensering

  Undersøkelsesmetode som gjør informasjonen i DNA molekylene våre om til noe som kan leses på en PC skjerm.
  Utviklet i 1977

Det humane genom på papir



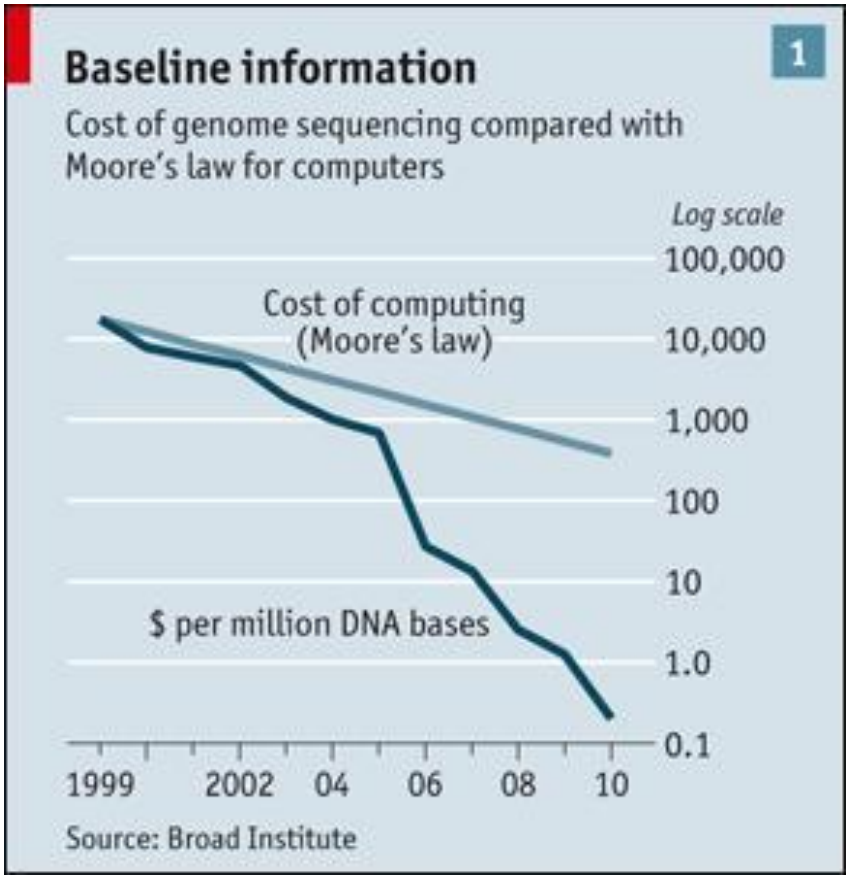
Mye vil skje i løpet av 10 år – jfr foregående tiår



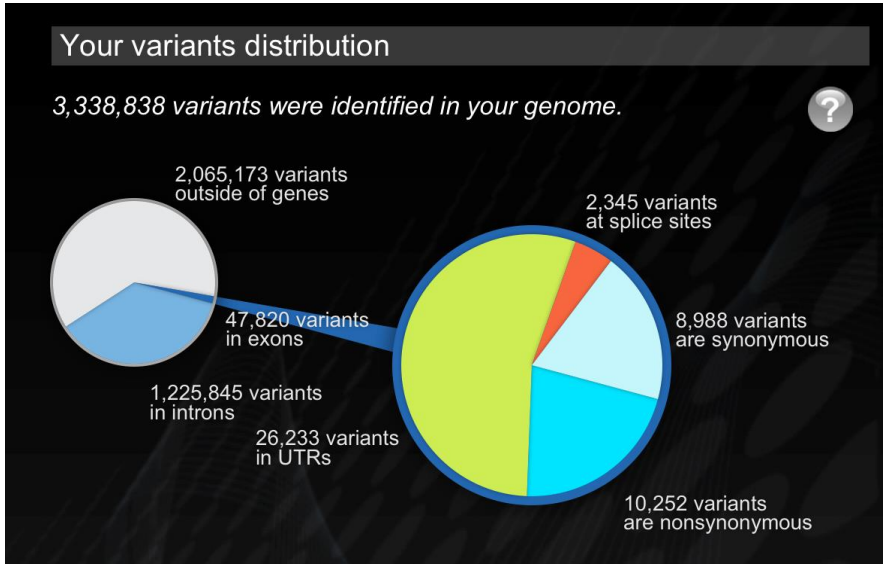
Ett humant genom

15 år

1 time



Genomsekvensering gir store datamengder om den enkelte



Mardis *Genome Medicine* 2010, **2**:84
http://genomemedicine.com/content/2/11/84

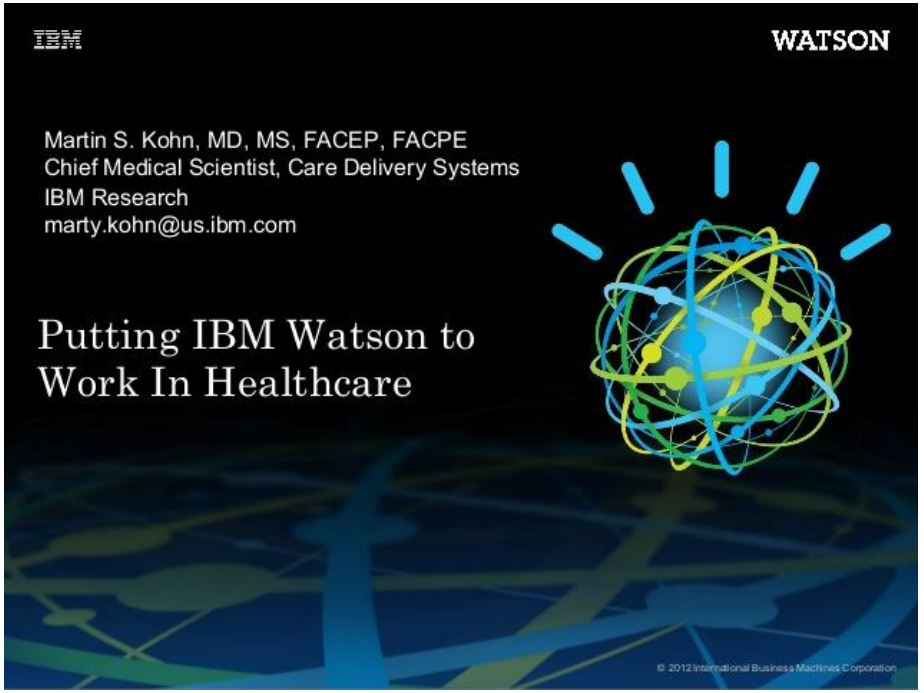
Genome Medicine

MUSINGS

The $1,000 genome, the $100,000 analysis?

Elaine R Mardis*

Utvikling av IKT beslutningsstøtte




IBM
SECOND MACHINE AGE
MACHINE TAKES OVER THINKING
Brynjulfsson & McAfee

Genomics england

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Genomics England, with the consent of participants and the support of the public, is creating a lasting legacy for patients, the NHS and the UK economy through the sequencing of 100,000 genomes: the 100,000 Genomes Project.

Genomics England was set up by the Department of Health to deliver the 100,000 Genomes Project. Initially the focus will be on rare disease, cancer and infectious disease. The project is currently in its pilot phase and will be completed by the end of 2017.

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Children could have DNA tested at birth

Jeremy Hunt wants Britain to become the first country in the world to routinely test children's entire DNA at birth to identify diseases

1 1 0 0 2 Email

Jeremy Hunt
Helseminister

Obama to Request Research Funding for Treatments Tailored to Patients' DNA

By ROBERT PEAR LAST UPDATED: JANUARY 25, 2015

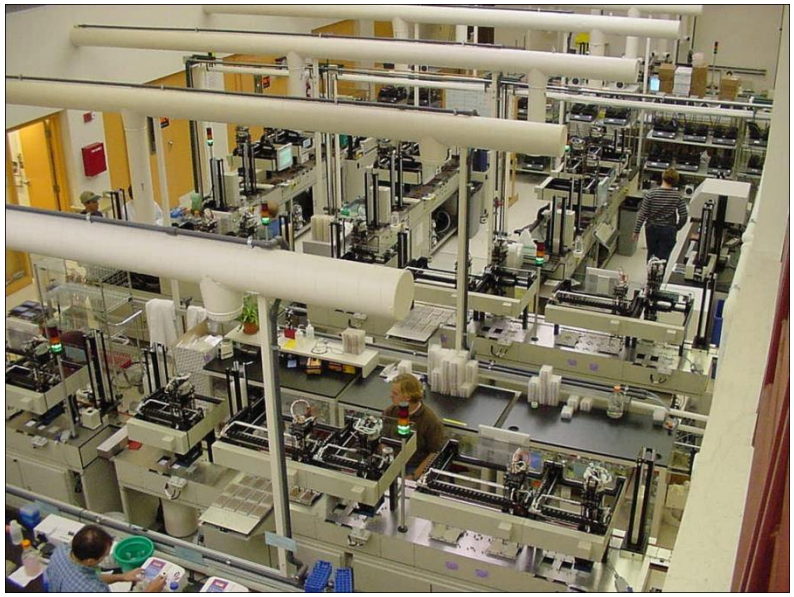
WASHINGTON — President Obama will seek hundreds of millions of dollars for a new initiative to develop medical treatments tailored to genetic and other characteristics of individual patients, administration officials say.

Video | Obama's State of the Union Address In his annual address before Congress, President Obama promised to confront growing economic inequality and outlined a list of actions he would take without Congressional approval.

The proposal, mentioned briefly in his State of the Union address, will be described in greater detail in his budget in the coming weeks. The effort is likely to receive support

Om 10 år vil mange i den norske befolkning vil ha fått kartlagt sitt genom

I en sekvenseringsfabrikk



Eller på kjøkkenet?

Om 10 år vil ”alle” enkeltgensykdommer ha funnet sin genetiske årsak

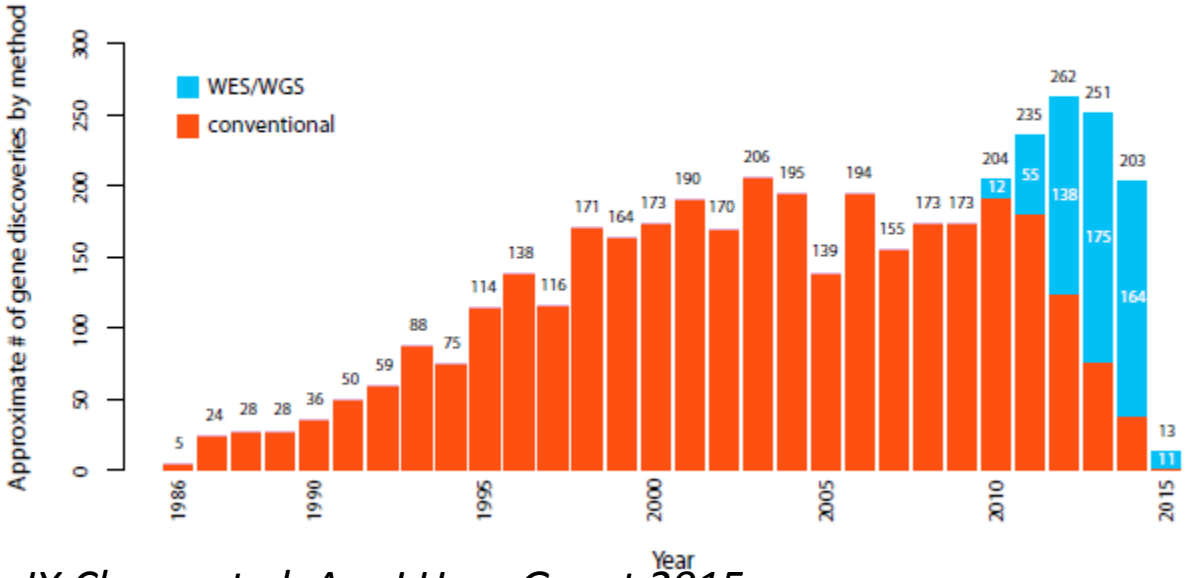
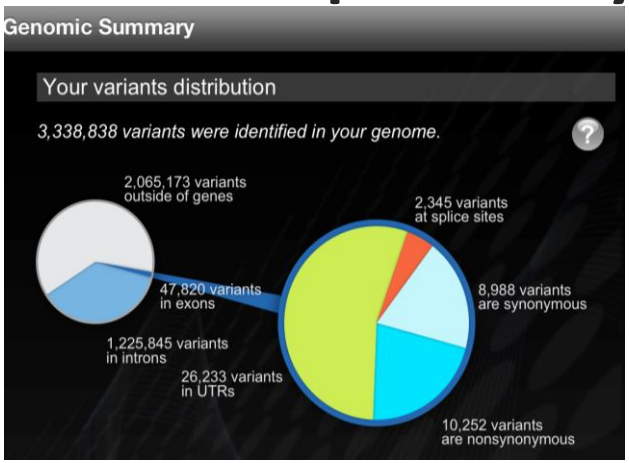


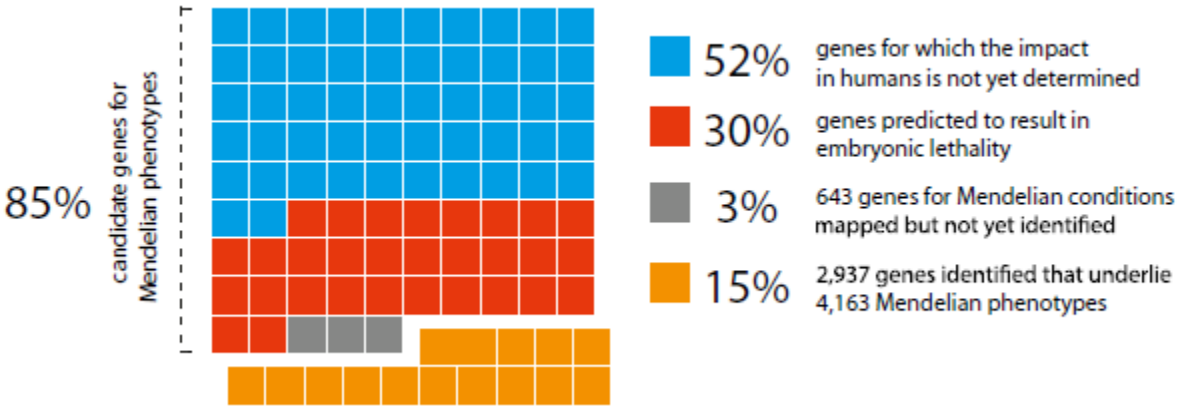
Figure 4. Approximate Number of Gene Discoveries Made by WES and WGS versus Conventional Approaches since 2010
Since the introduction of WES and WGS in 2010, the pace of discovery of genes implicated in Mendelian phenotypes per year has increased substantially, and the proportion of discoveries made by WES or WGS (blue) versus conventional approaches (red) has steadily increased (see Supplemental Material and Methods for a detailed description of the analysis). Since 2013, WES and WGS have discovered nearly three times as many genes as conventional approaches.

JX Chong et el. Am J Hum Genet 2015

Om 10 år vi kunne høste mye mer medisinsk nytte ut av økt kunnskap om betydningen av genetisk variasjon



Vi har kun ”medisinsk nyttig” kunnskap om den kodende del av genomet (1-1,5%)



>50% av gener har ukjent betydning

JX Chong et el. Am J Hum Genet 2015

Totalt er det i dag < 1% av variasjonen i genomet som er klinisk nyttig

Fra anekdotiske historier til dagligdags helsehjelp

Om 10 år vil genomsekvensering være hverdagslig
i det norske helsevesen

Genomsekvensering er på full fart inn i diagnostikk av arvelige sykdommer allerede i dag og vil være rutine om 10 år

Gilissen *et al. Genome Biology* 2011, **12**:228
http://genomebiology.com/2011/12/9/228

Genome Biology

REVIEW

Unlocking Mendelian disease using exome sequencing

Christian Gilissen*, Alexander Hoischen, Han G Brunner and Joris A Veltman

Abstract

Exome sequencing is revolutionizing Mendelian disease gene identification. This results in improved clinical diagnosis, more accurate genotype-phenotype correlations and new insights into the role of rare genomic variation in disease.

Kreft – en sykdom som skyldes genfeil

Genetic Gamble

New Approaches to Fighting Cancer

PART ONE
A Race to Leukemia's Source

PART TWO
Promise and Heartbreak

In Treatment for Leukemia, Glimpses of the Future

Second Chance: Lukas Wartman, a leukemia doctor and researcher, developed the disease himself. As he faced death, his colleagues sequenced his cancer genome. The result was a totally unexpected treatment.

By GINA KOLATA

Mikrober har også DNA

In a First, Test of DNA Finds Root of Illness

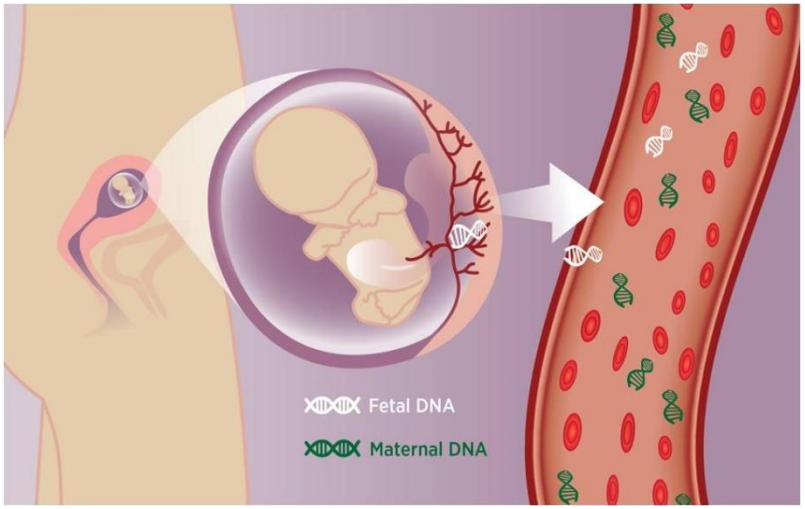
By CARL ZIMMER LAST UPDATED: JUNE 4, 2014

Joshua Osborn, 14, lay in a coma at American Family Children's Hospital in Madison, Wis. For weeks his brain had been swelling with fluid, and a battery of tests had failed to reveal the cause.

Joshua Osborn outside his home in Cottage Grove, Wis., with his father, Clark Osborn. Joshua suffered from swelling in the brain, but tests, a spinal tap and a biopsy were inconclusive. JOHN MANIACI.

The doctors told his parents, Clark and Julie, that they wanted to run one more test with an experimental new technology. Scientists would search Joshua's cerebrospinal fluid for pieces of DNA. Some of them might belong to the pathogen causing his

Risikofri fosterdiagnostikk



0,5% risiko for uønsket abort

Fritt sirkulerende DNA fra foster kan gjenfinnes i mors blod og kan testes mtp arvelige sykdommer

Om 10 år vil vi ha begynt å høste nytteverdien av genetiske oppdagelser i form av nye ”genspesifikke” medisiner

Vil genterapi omsider bli en suksess?

Om 10 år vil undersøkelse av det dynamiske genomet være på vei inn i medisinsk rutine

- Kartlegging av det ”statiske” genomet er på full fart inn i helsevesenet i dag
  - Gjør det mulig å identifisere faktorer som gir arvelige sykdommer
- Genomet har også en dynamisk side – epigenetikk, genekspresjon etc
  - Per i dag er teknologien for umoden og kunnskapsgrunnlaget for tynt til at undersøkelse av dynamiske endringer har blitt tatt i bruk i stor grad i helsevesenet
  - Om 10 år er det sannsynlig at dette har endret seg
- Dette vil kunne ha stor betydning for undersøkelse og behandling av miljøindusert sykdom, livsstilssykdommer med mer.

Men husk

”The process for establishing appropriate policies for implementation of new scientific findings can be frustratingly slow. The median time lag between the earliest publication of a medical discovery and its implementation was 24 years”

Francis Collins NIH Director

og

Det er svært at spå, især om fremtiden

Takk for oppmerksomheten!