

Welcome

GENETICS, ETHICS AND CLINICAL TRANSLATION

Summer Institute, June 22 – 25, 2010

Hotel Zuiderduin, Egmond aan Zee, The Netherlands

This four-day intensive course is designed for professionals actively involved in biobanking who want to learn more about the **ethical, legal and societal** implications of the translation of epidemiological results from genetic and genomic research to clinical practice.

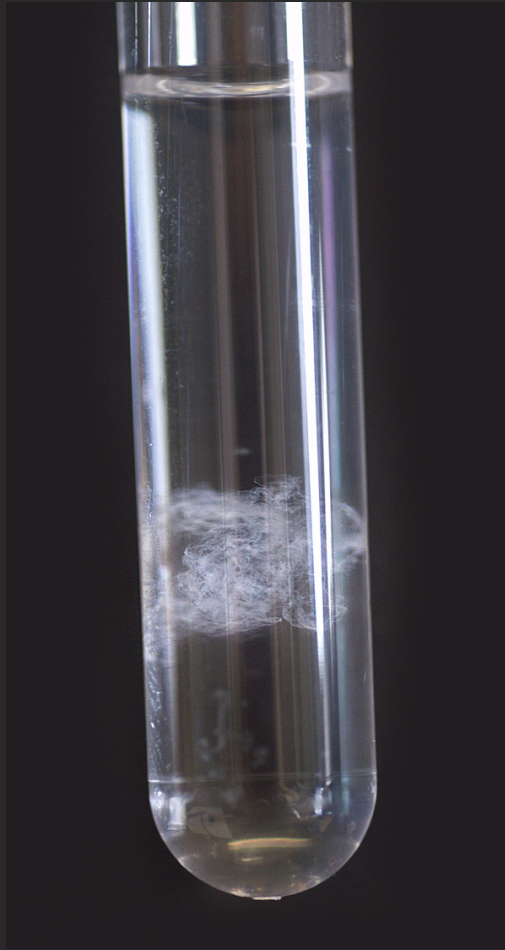
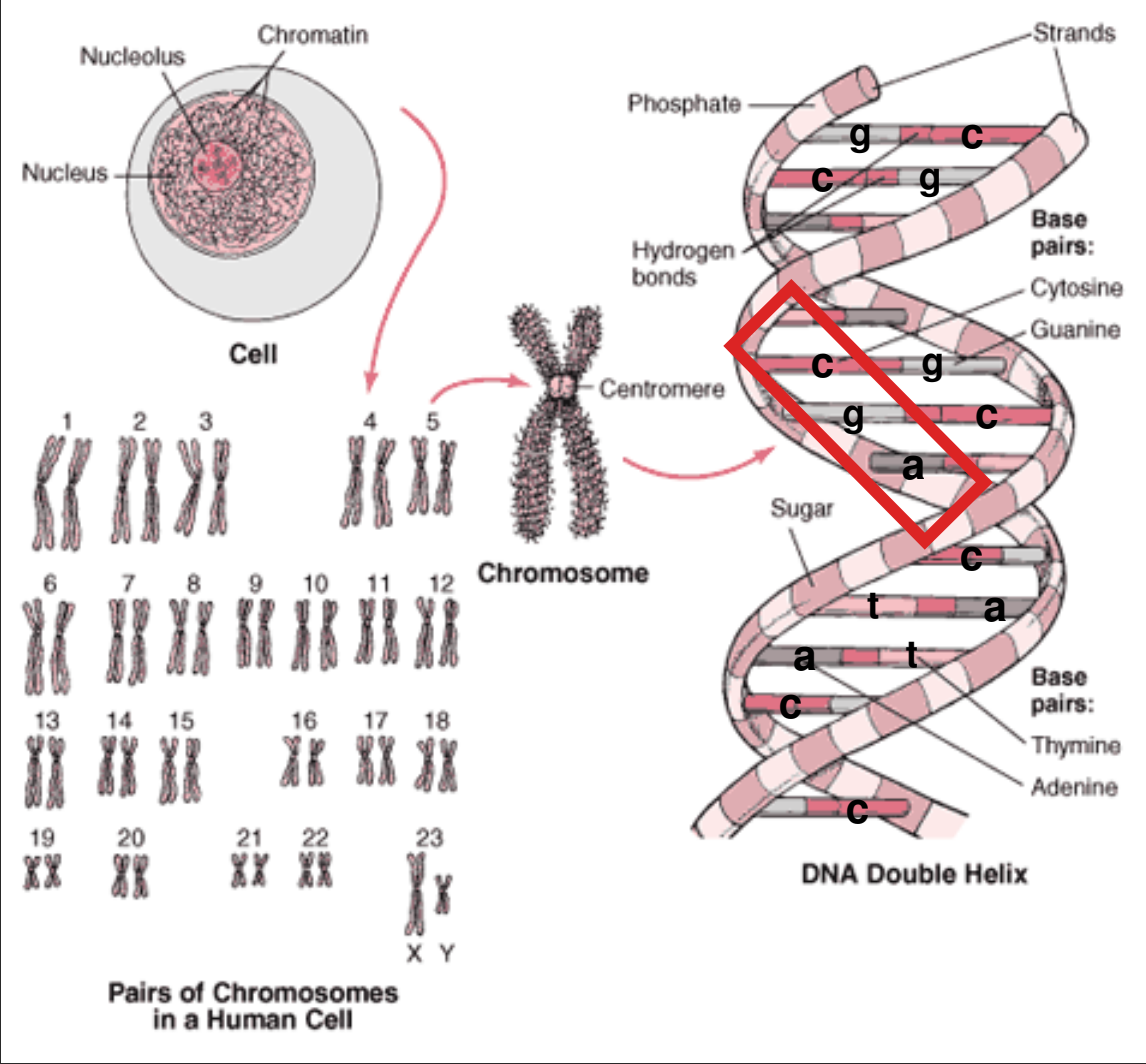
The Summer Institute will offer insight about issues and solutions applicable to the field, and will give participants the opportunity to actively interact with the lecturers through debates and working group sessions. The course will total 17 hours of teaching and work group sessions.

Three main topics will be covered

- Introduction to translational research and ethical, legal and societal issues in biobanking
- The implications of genetic testing, personal genome and personalized medicine
- Ways to handle the interface with stakeholders and society

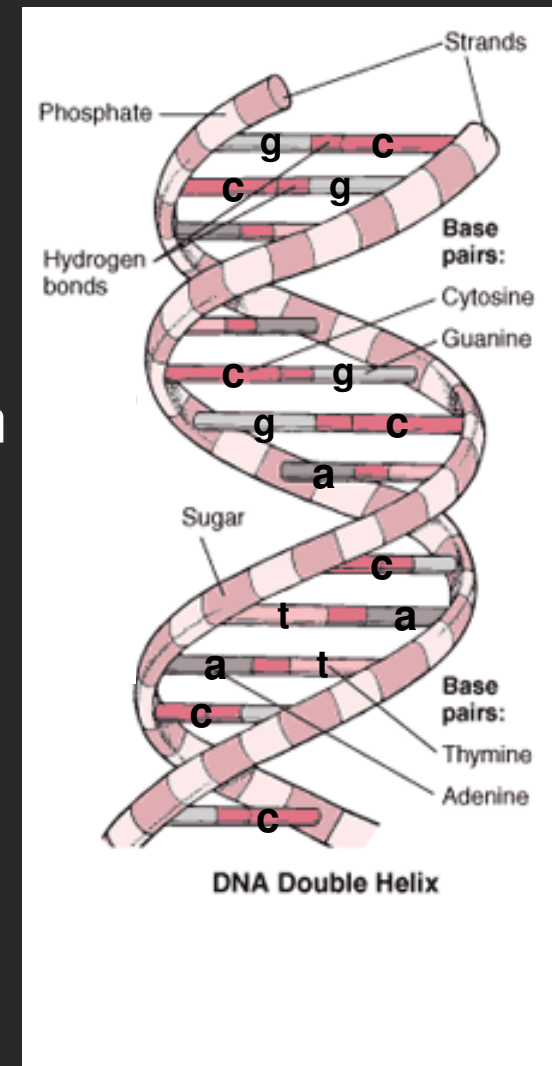


DNA

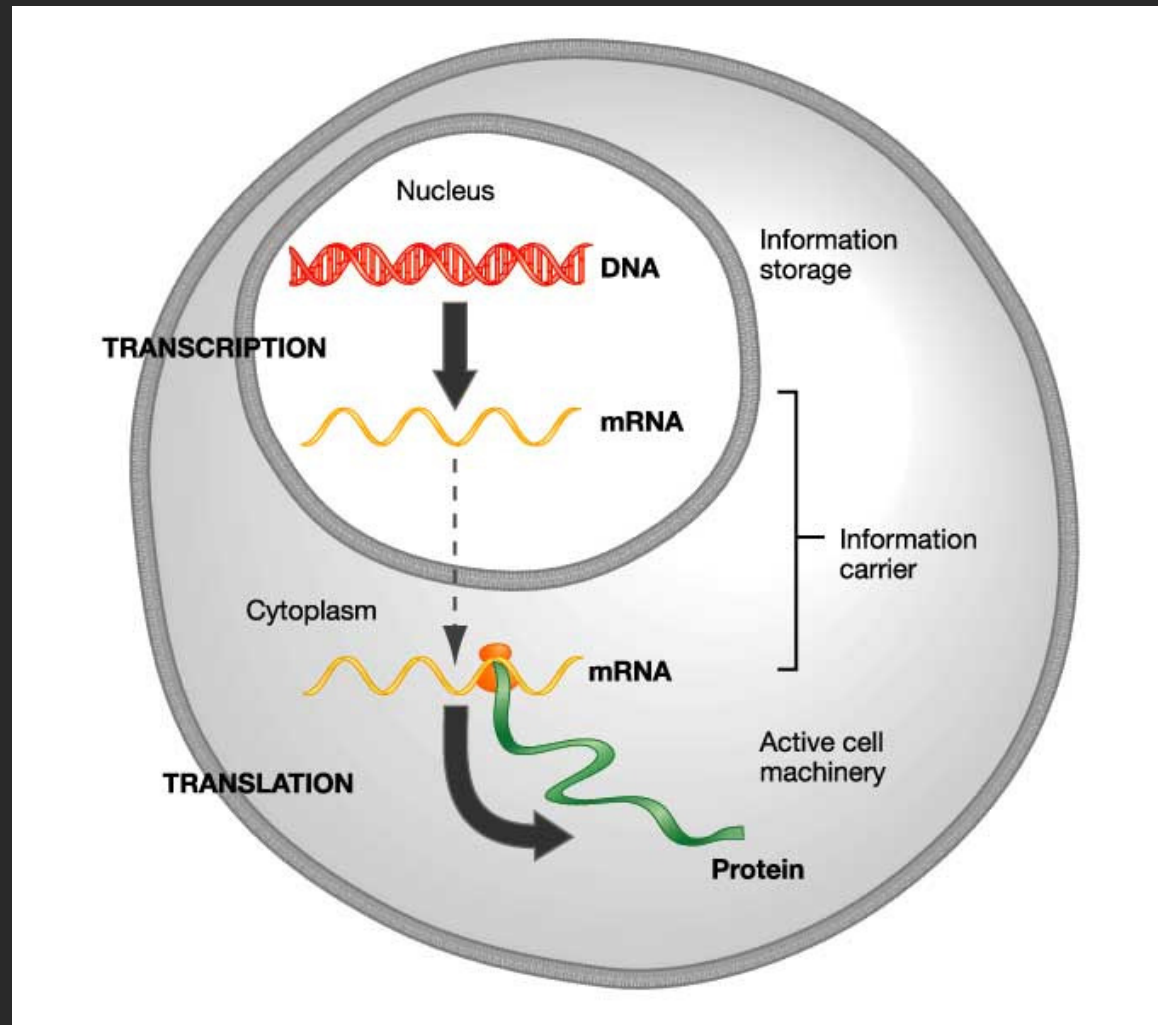


Gene

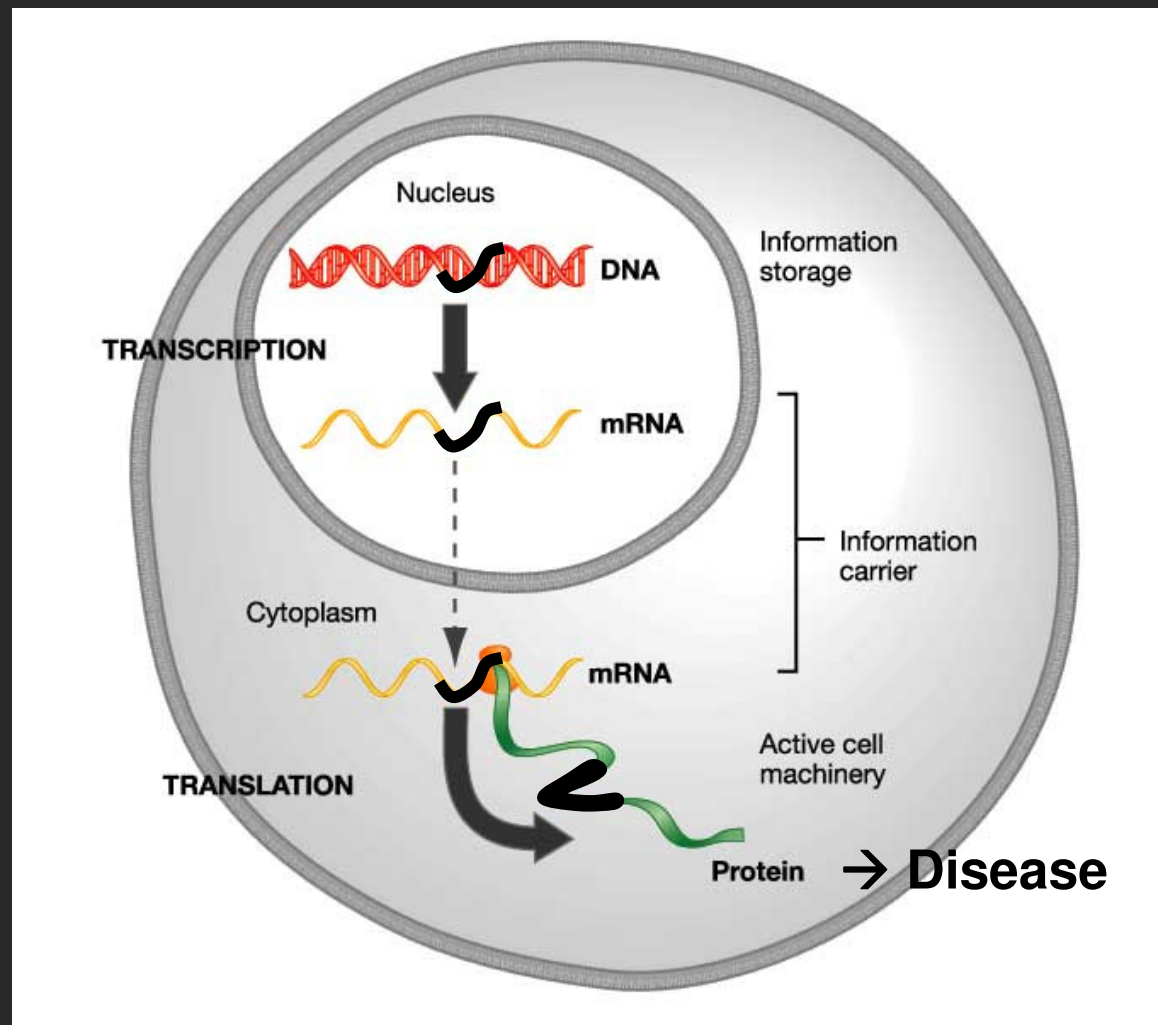
- Basic unit of heredity
- **Sequence of DNA** that encodes a protein
- Human DNA: 3.000.000.000 basepairs
- Human DNA: ~ 30,000 genes
- Average length: ~ 30,000 base pairs
- Genes make up < 1% of DNA



From gene to protein



From gene to protein **to disease**



What can go wrong at DNA level?

Types of Markers

- Blood Group Markers
- Restriction Fragment Length Polymorphisms (RFLPs)
- Insertions/Deletions (Indels)
- Variable Number of Tandem Repeats (VNTRs)
- Microsatellites (Short Tandem Repeat Polymorphisms [STRPs])
- Single Nucleotide Polymorphisms (SNPs)
- Copy Number Variants (CNVs)
- Epigenetic Changes
- Whole Genome Sequencing

← currently of major interest

Genetic association study

Persons with disease

aa aa aa aa aa aa at at at tt

$$a / t = 15 / 5 = 3$$

Persons without disease

aa at at at tt tt tt tt tt tt tt

$$a / t = 5 / 15 = 0.33$$

Conclusion: allele 'a' of snp x is associated to disease risk (OR = 9)

Next scientific questions:

- Where in DNA is snp x located?
- If in gene, what is the function of the protein that is coded by the gene?

At same time commercial interest:

- Can we make a test of it?
- (is not real question: answer is no, but they just do it)

Genetic associations are ...

- ... investigated in large-scale biobanks
- the basis for genetic tests both
 - in health care practice and ...
 - commercially offered tests

Tuesday 22nd June 2010

Session 1 - Introduction to translational research and ELSI issues in biobanking	
Moderator: Cecile Janssens, Erasmus University Medical Center	
09.00	Welcome
09.15	Prospective/retrospective biobanking and translational research Jennifer Harris, Norwegian Institute of Public Health, Norway
10.00	Coffee break
10.15	Ethical, Legal and Societal issues in biobanking Bartha M Knoppers, McGill University, Canada
11.15	Coffee break
11.30	Biobanking and public health: uses and ELSI implications Marta Gwinn, National Office of Public Health Genomics, Centers for Disease Control and Prevention, USA
12.15	Lunch
13.30	Working group session: Case study Session chair: Bartha M Knoppers, McGill University
14.30	Coffee break
14.45	Expectations about the Future of Personal Genomics: Markets, Investors, Regulators and Users Richard Tutton, Lancaster University, UK
15.30 – 16.00	Discussion in plenum and wrap-up

Wednesday 23rd June 2010

Session 2 - Towards translation: genetic testing, personal genome and personalized medicine	
Moderator: Bartha M Knoppers, McGill University, Canada	
09.00	Introduction to genetic testing and pharmacogenomics Cecile Janssens, Erasmus University Medical Center, The Netherlands
09.45	Coffee break
10.00	Genetic screening: policy, organization and update Martina Cornel, VU University Medical Centre, The Netherlands
10.45	Coffee break
11.00	Psychological and behavioural implications of genetic testing Lidewij Henneman, VU University Medical Centre Amsterdam, The Netherlands
11.45	Discussion in plenum
12.15	Lunch
13.30	Working group session: Case study Session chair: Cecile Janssens, Erasmus University Medical Center
14.30	Coffee break
14.45	Ethical, legal and societal implications of genetic testing Anne-Marie Tassé, McGill University, Canada
15.30 – 16.00	Discussion in plenum and wrap-up

Thursday 24th June 2010

Session 2 continues: Towards translation: genetic testing, personal genomes and personalized medicine	
Moderator: Isabelle Budin Ljøsne, Norwegian Institute of Public Health, Norway	
09.00	Introduction to commercial and recreational testing Cecile Janssens, Erasmus University Medical Center, The Netherlands
09.45	Coffee break
10.00	Ethical aspects of commercial and recreational testing Heidi Howard, Centre for Biomedical Ethics and Law, Belgium
10.45	Coffee break
11.00	Policy aspects of commercial and recreational testing Christine Patch, Guys Hospital, UK
11.45	Discussion in plenum
12.15	Lunch
	Afternoon: Free time

Friday 25th June 2010

Session 3 - Engaging stakeholders and society	
Moderator: Jennifer Harris, Norwegian Institute of Public Health	
09.00	Introduction to interactions and strategies Jacqueline Broerse, VU University Amsterdam, The Netherlands
09.45	Coffee break
10.00	Are the respect for privacy and open source compatible? Yann Joly, McGill University, Canada
10.45	Coffee break
11.00	Working group session with tutor: How to communicate scientific results to stakeholders? Session chair: Denise Avar, McGill University, Canada
12.00	Lunch
13.00	Movie/ role playing Session chair: Frans van Dam, CSG, The Netherlands
15.00 – 15.15	Wrap-up and evaluation