

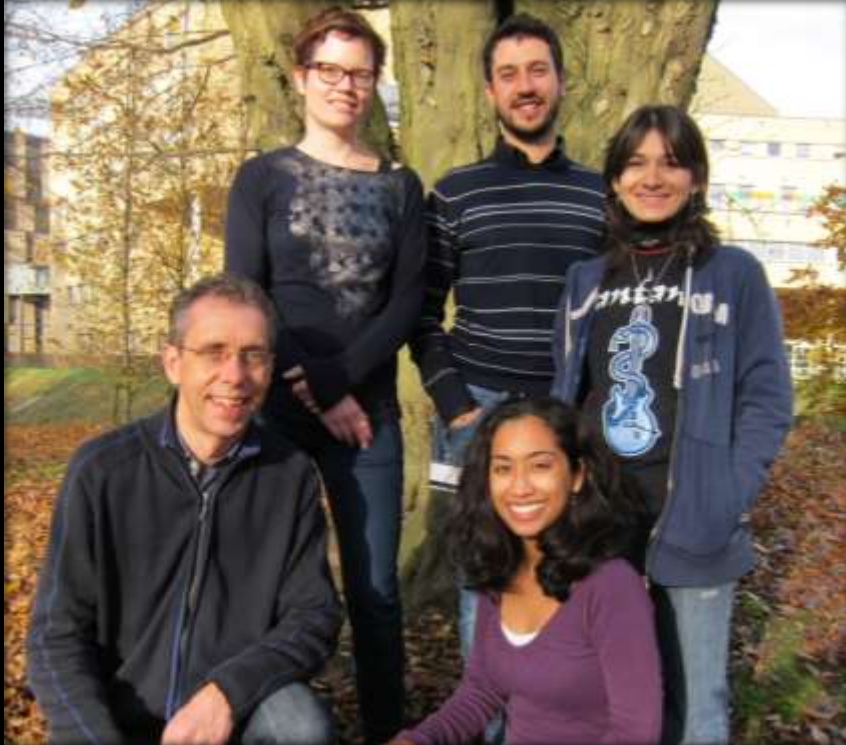
Genetische studies DCMD

Prof. dr. Frans P.M. Cremers

Afd. Genetica, RadboudUMC

7-2-2015; Nijmegen, patiëntendag DCMD

Onderzoeksteam



Genetica Nijmegen:

Silvia Albert

Riccardo Sangermano

Anke den Engelsman-van Dijk

Angelique Ramlal

Frans Cremers

Oogheelkunde Nijmegen:

Mahesh Duvvary

Nicole Saksens

Anneke den Hollander

Carel Hoyng

Leiden: Camiel Boon

Financiering

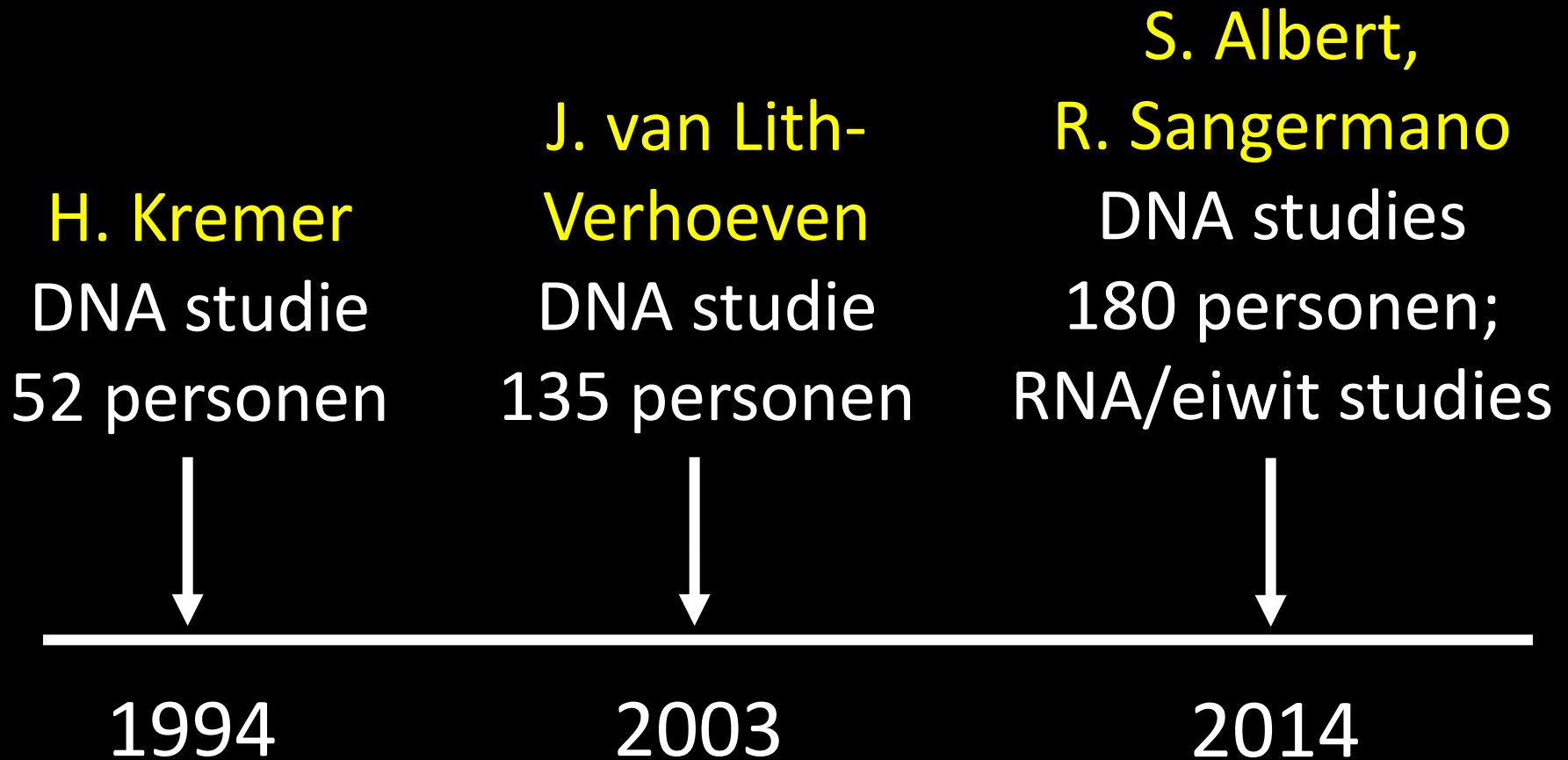


EU-ITN

CYMD stichting

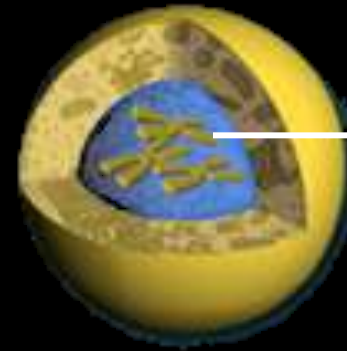
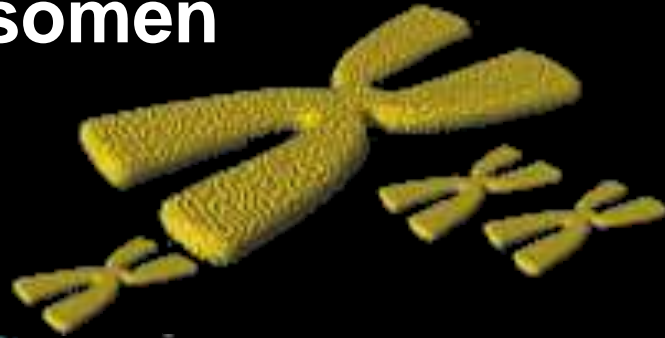


Genetische studies DCMD



Genetische informatie zit in de kern van de cel

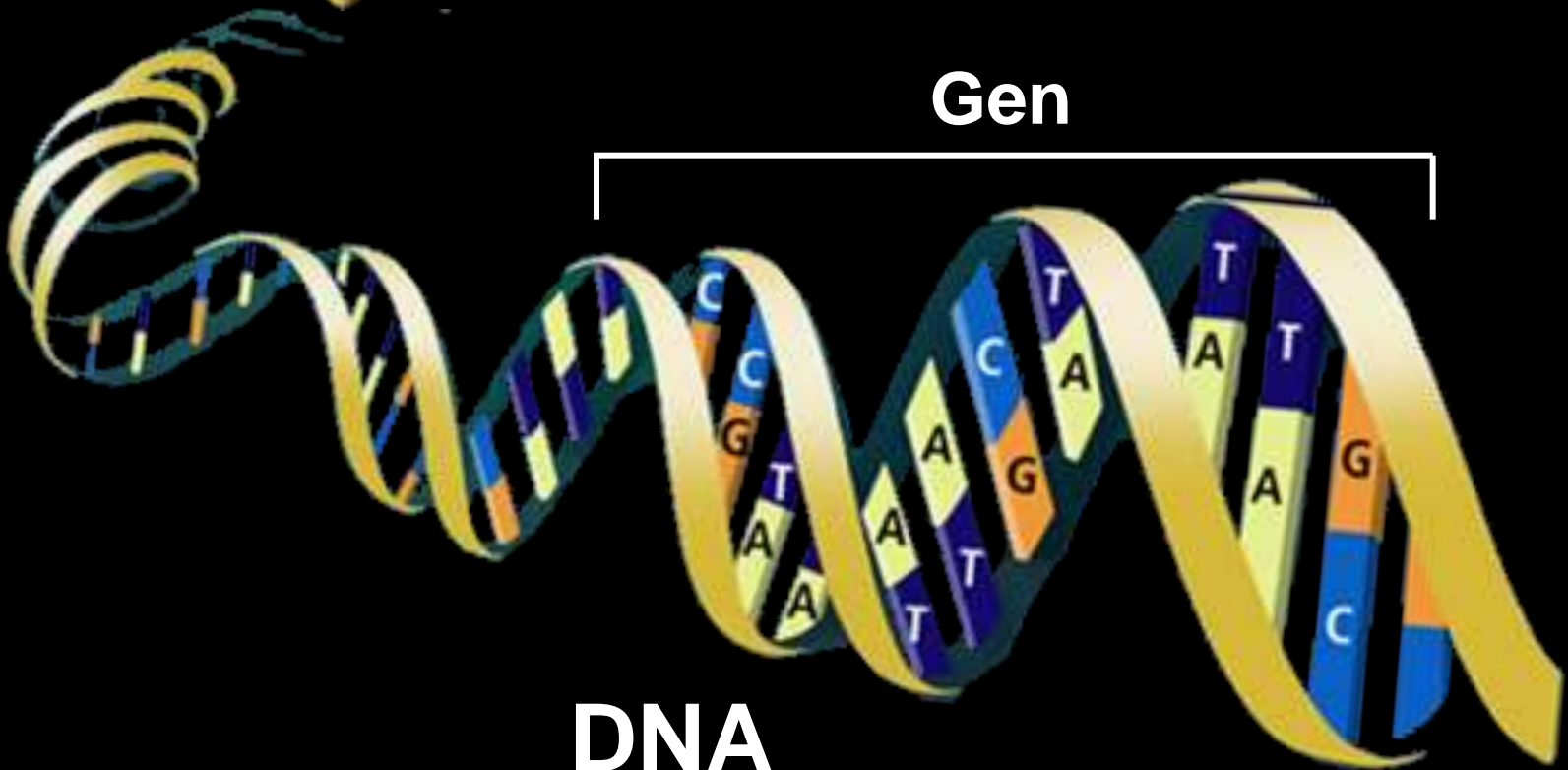
Chromosomen



Kern

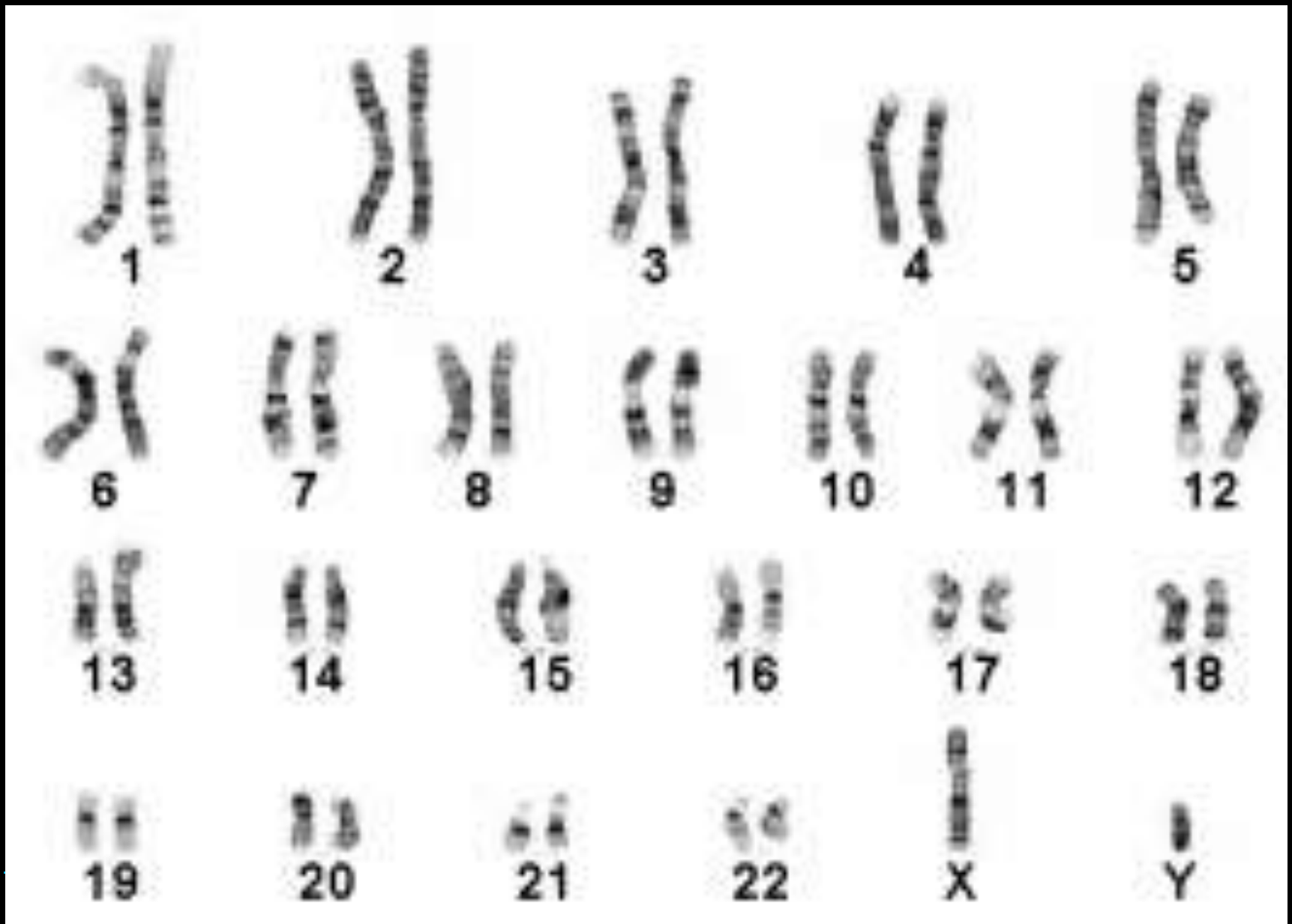
Cel

Gen



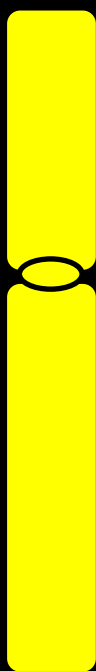
DNA

Iedere kern bevat 23 paar chromosomen



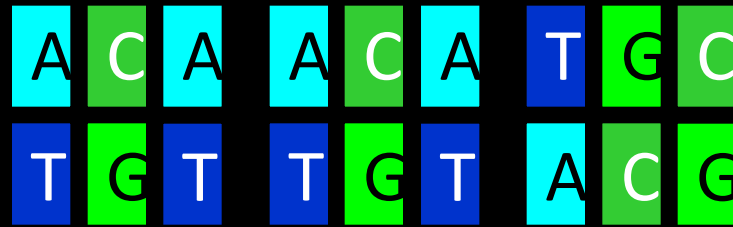
**Chromosoom
van vader**

**Chromosoom
van moeder**

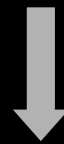


1000 genen

DNA



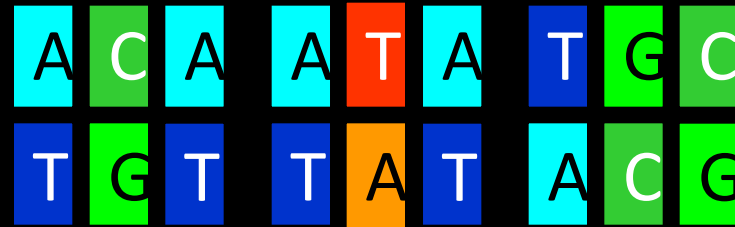
RNA



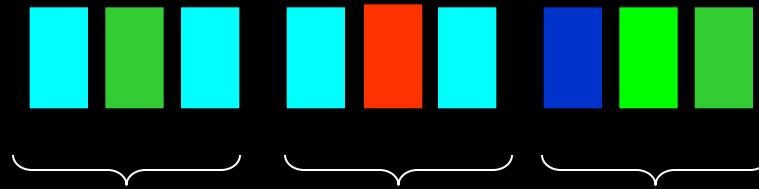
Eiwit



DNA



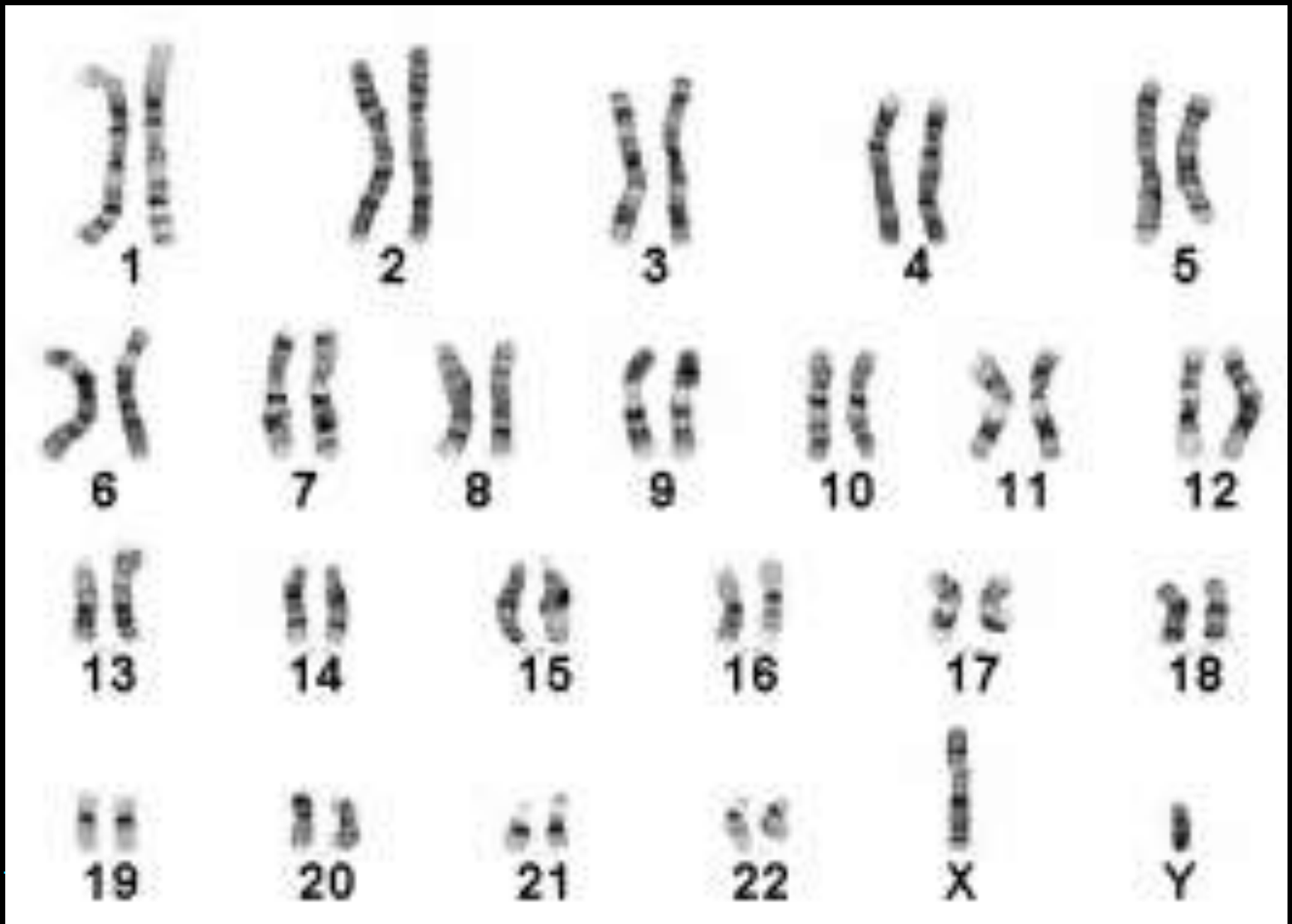
RNA



Defect
eiwit



Iedere kern bevat 23 paar chromosomen



In menselijk DNA

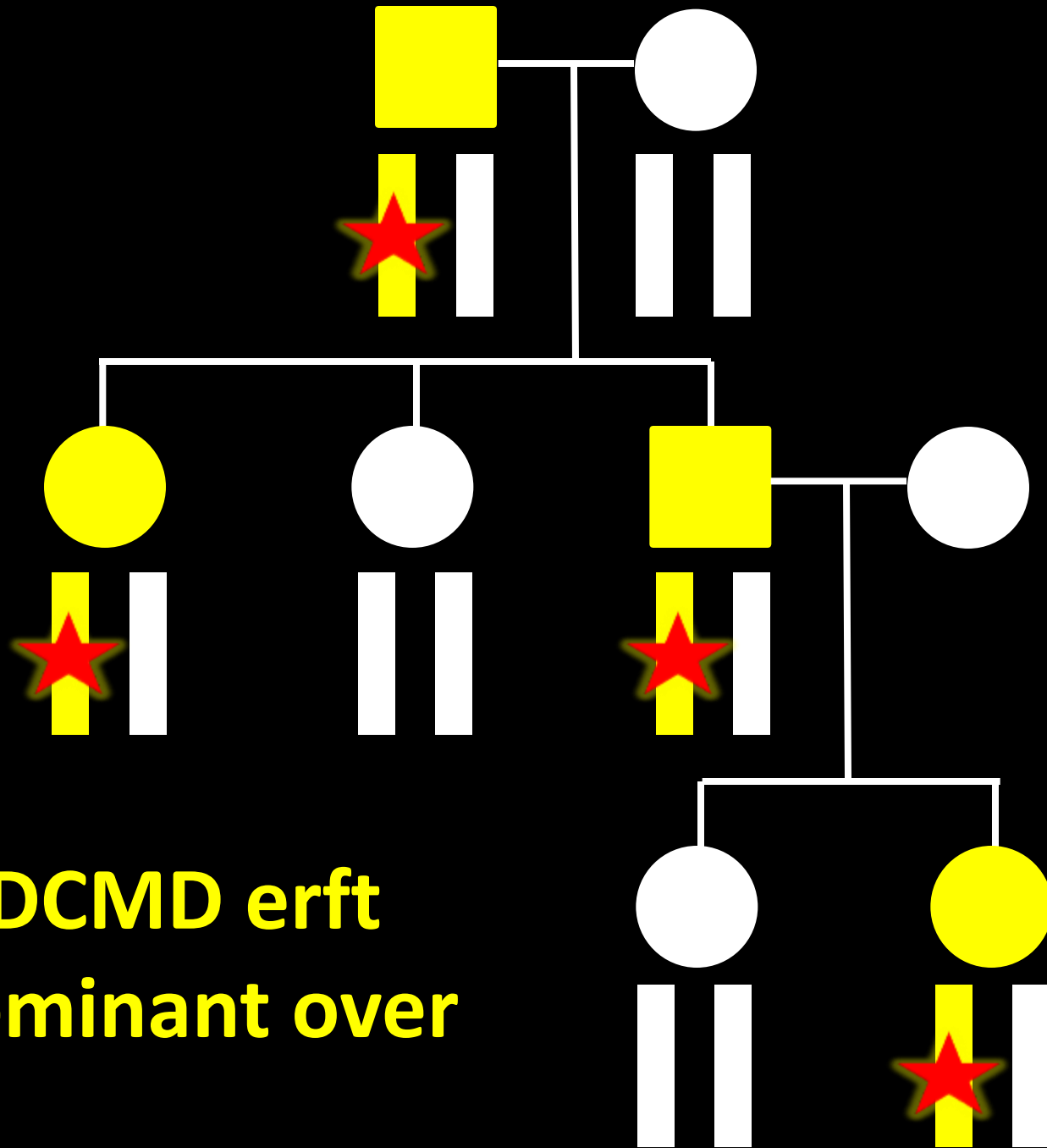
0.1% = 3 miljoen bp verschillen



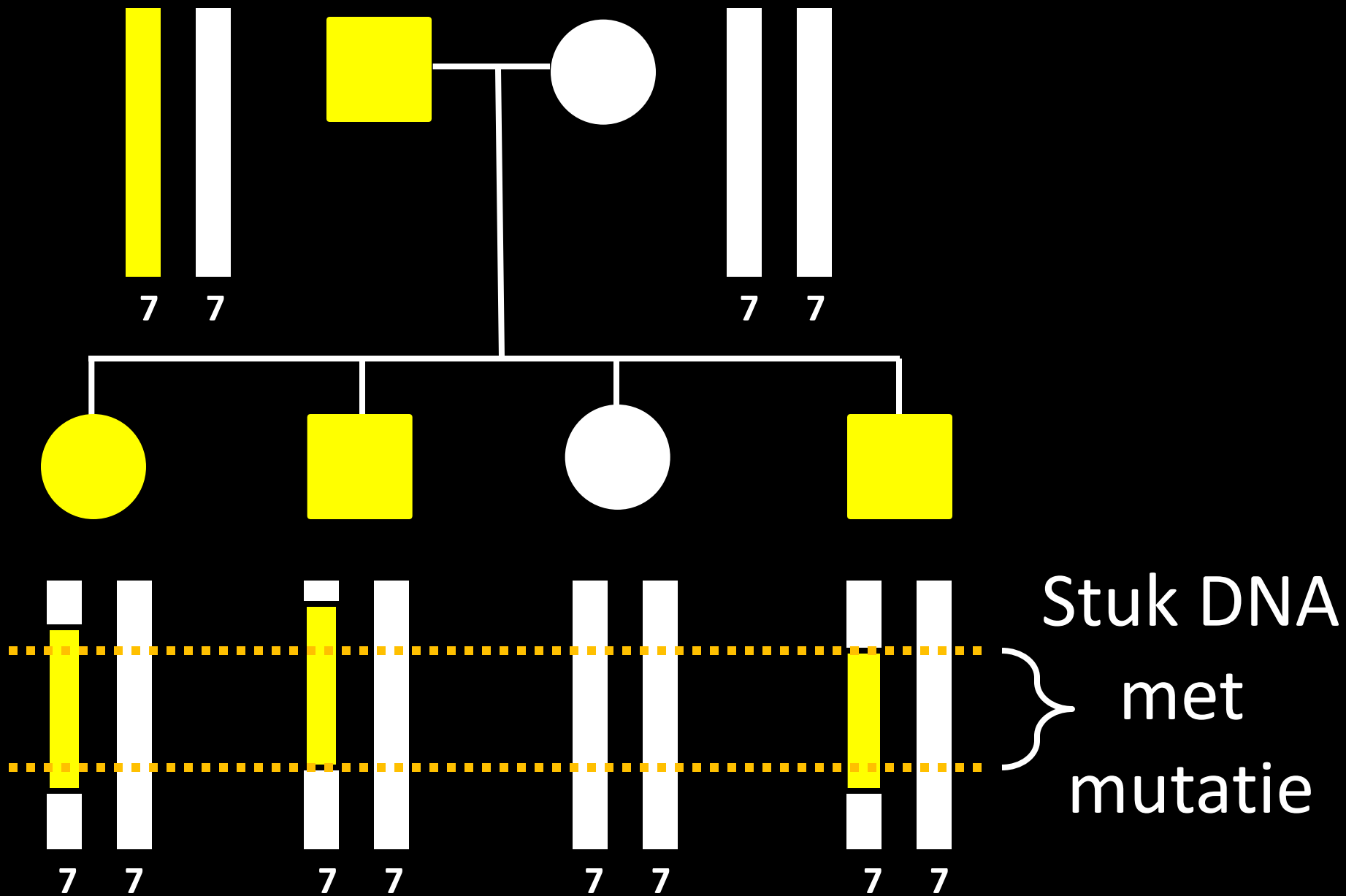
agaagcctcctaggggttgcaagcct**C**cttggctaattggttgggct

agaagcctcctaggggttgcaagcct**G**cttggctattggttgggct





**DCMD erft
dominant over**

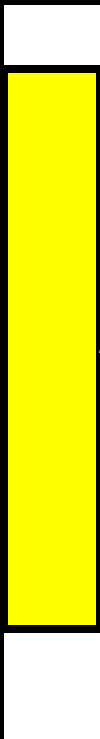


Chr. 7



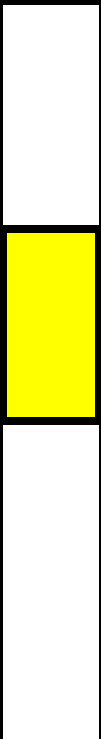
~2.000 genen

1994



**~250
genen**

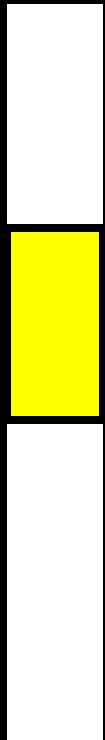
2003



**~50
genen**

**Hannie Kremer
Janneke van Lith**

2003



~50

genen

2010: Sequentie-analyse alle 50 genen met nieuwe methode.

Anneke den Hollander

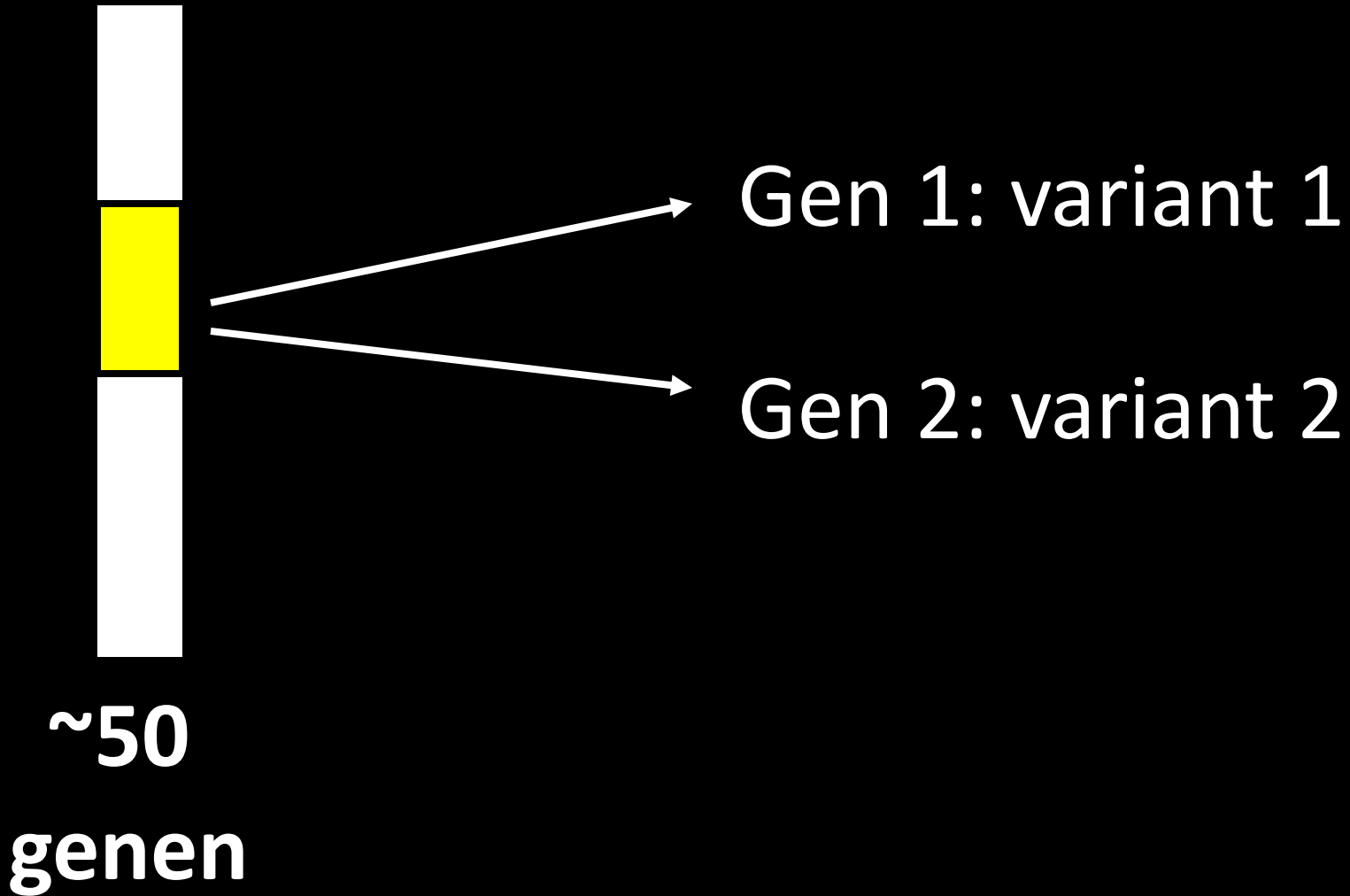
2014: Sequentie-analyse alle genen met meer complete methode → analyse is net klaar

Riccardo Sangermano

Silvia Albert

(in samenwerking met Carlo Rivolta, Lausanne, Zwitserland)

Kandidaat-varianten:



Studies in komende maanden:

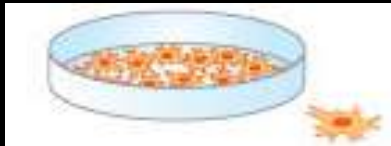
- **Analyse nieuwe mutatie(s) in alle personen van grote DCMD familie**
- **Analyse gemuteerde genen in 5 andere kleine families met DCMD**
- **Indien oorzaak niet gevonden:
analyse RNA**

Kunstmatige netvliescellen

Huidbiopt



Fibroblasten



Stamcel



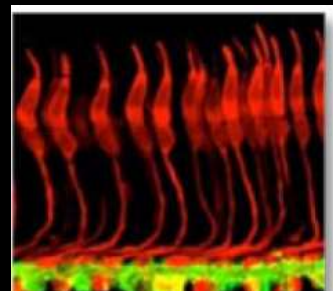
RNA sequentie-
analyse



RNA isolatie



Netvliescellen



Eiwit-studies



Communicatie

Facebook: Blindness Genetics
workgroup; google “facebook vision
institute nijmegen”

[https://www.facebook.com/pages/
Vision-Institute-Nijmegen/328133970713796](https://www.facebook.com/pages/Vision-Institute-Nijmegen/328133970713796)

Website @ RadboudUMC:

[https://www.radboudumc.nl/Research/Organisationofresearch/
Departments/HumanGenetics/Pages/BlindnessGenetics.
aspx](https://www.radboudumc.nl/Research/Organisationofresearch/Departments/HumanGenetics/Pages/BlindnessGenetics.aspx)