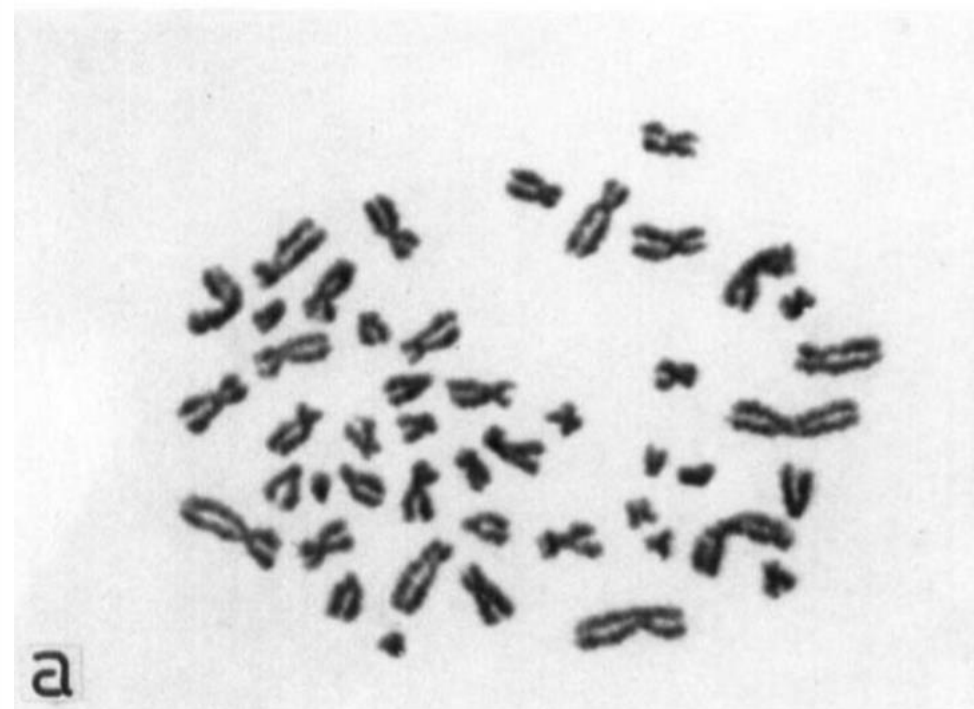


Dokter & Onderzoeker in Nederland & United Kingdom

Fleur S. van Dijk

Klinische genetica

- Sinds 1987 erkend als medisch specialisme
- Jongste geneeskundig specialisme
- In elk academisch ziekenhuis is een afdeling klinische genetica, vanuit deze afdeling wordt poli gedaan in perifere ziekenhuizen in de regio
- Oncogenetica, Cardiogenetica, Dysmorfologie
- Zelf gespecialiseerd in erfelijke bindweefselaandoeningen



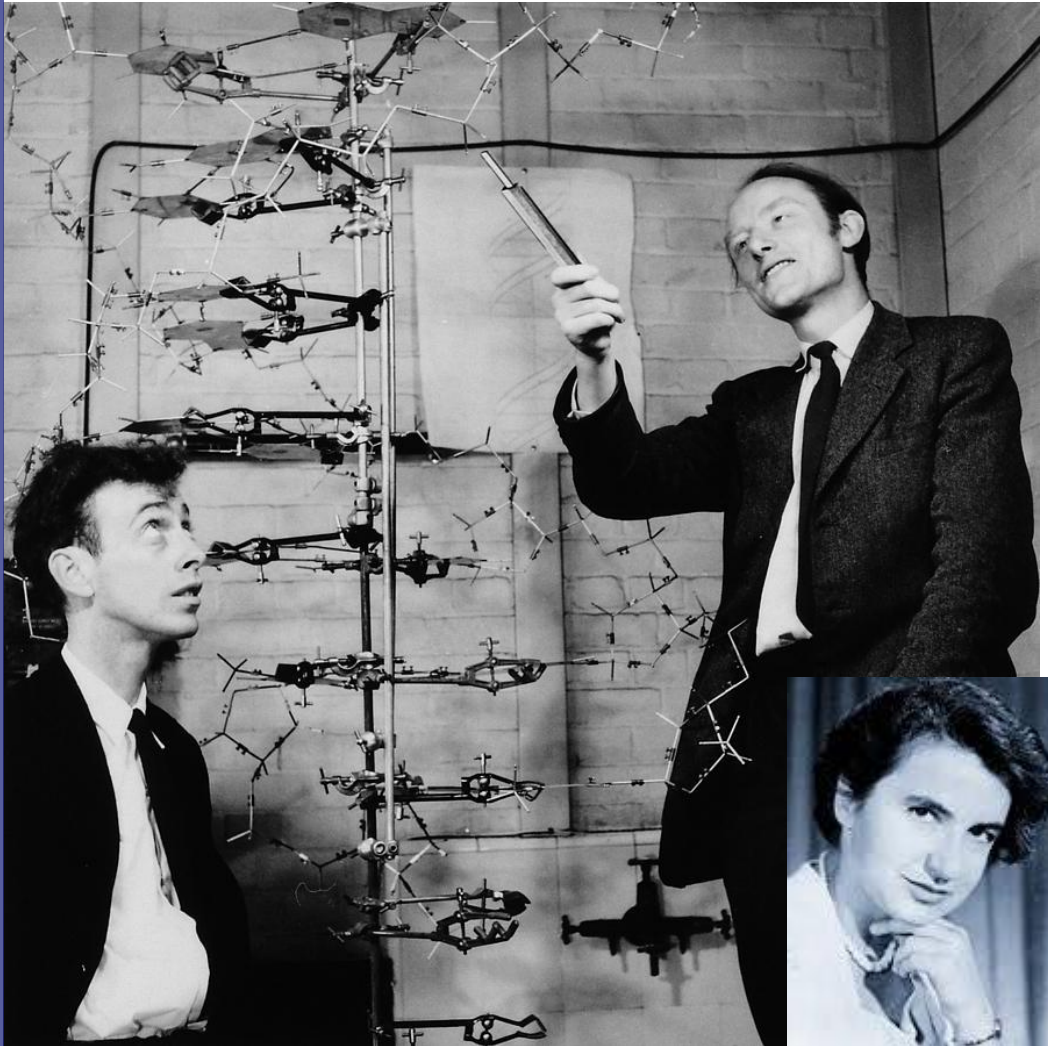
THE CHROMOSOME NUMBER OF MAN

By *JOE HIN TJIO* and *ALBERT LEVAN*

ESTACION EXPERIMENTAL DE AULA DEI, ZARAGOZA, SPAIN, AND CANCER CHROMOSOME
LABORATORY, INSTITUTE OF GENETICS, LUND, SWEDEN

- In 1956 werd duidelijk dat mensen in elke celkern 46 chromosomen hebben.
- Eerste numerieke afwijkingen: Down syndrome (trisomy 21, 1959), Edwards (trisomy 18), Patau (trisomy 13), Klinefelter, Turner,
- Banderings technieken om ongebalanceerde translocaties te ontdekken werden pas later beschikbaar (Q-banding (eind Jaren 60), G-banding (begin jaren 70)

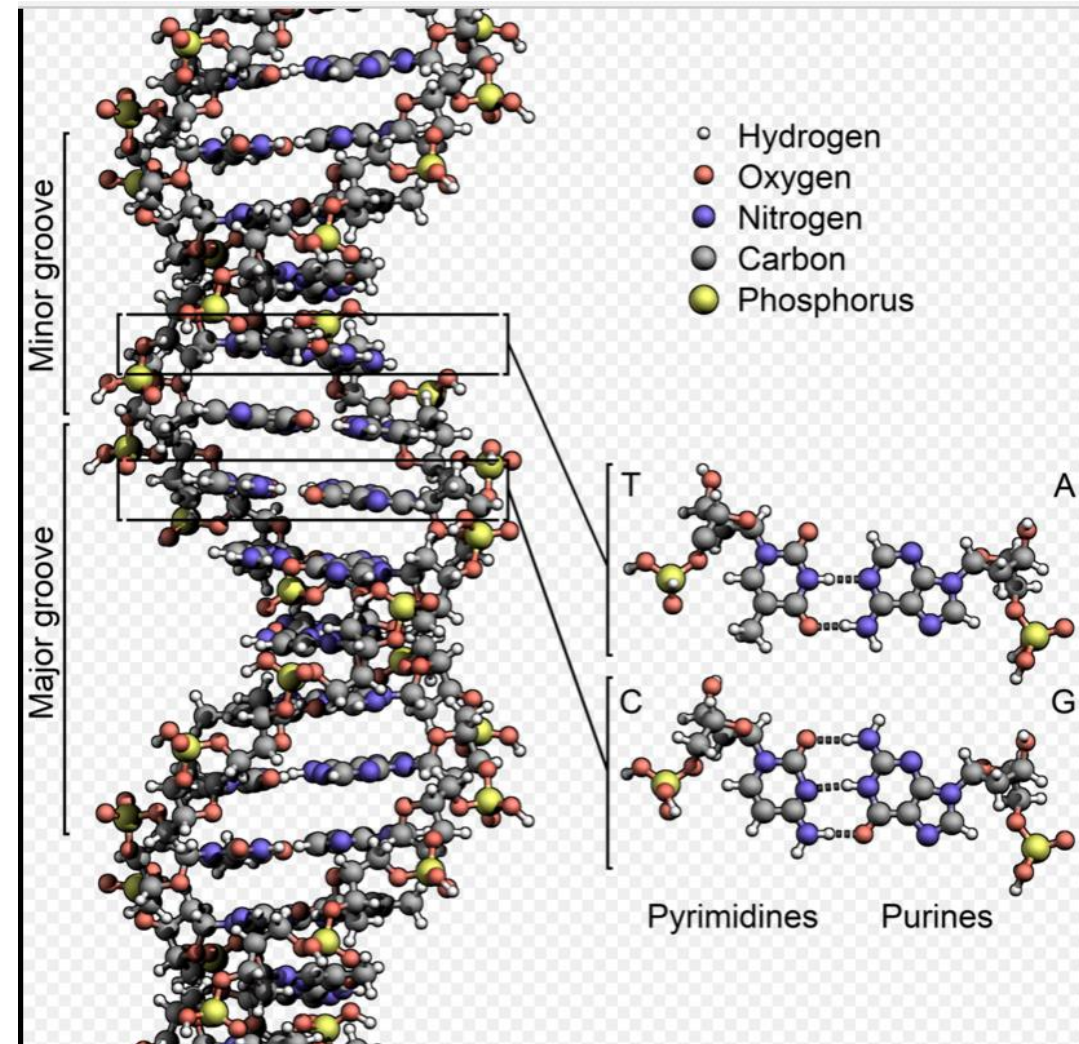
Deoxyribonucleic acid (DNA): structuur 1953



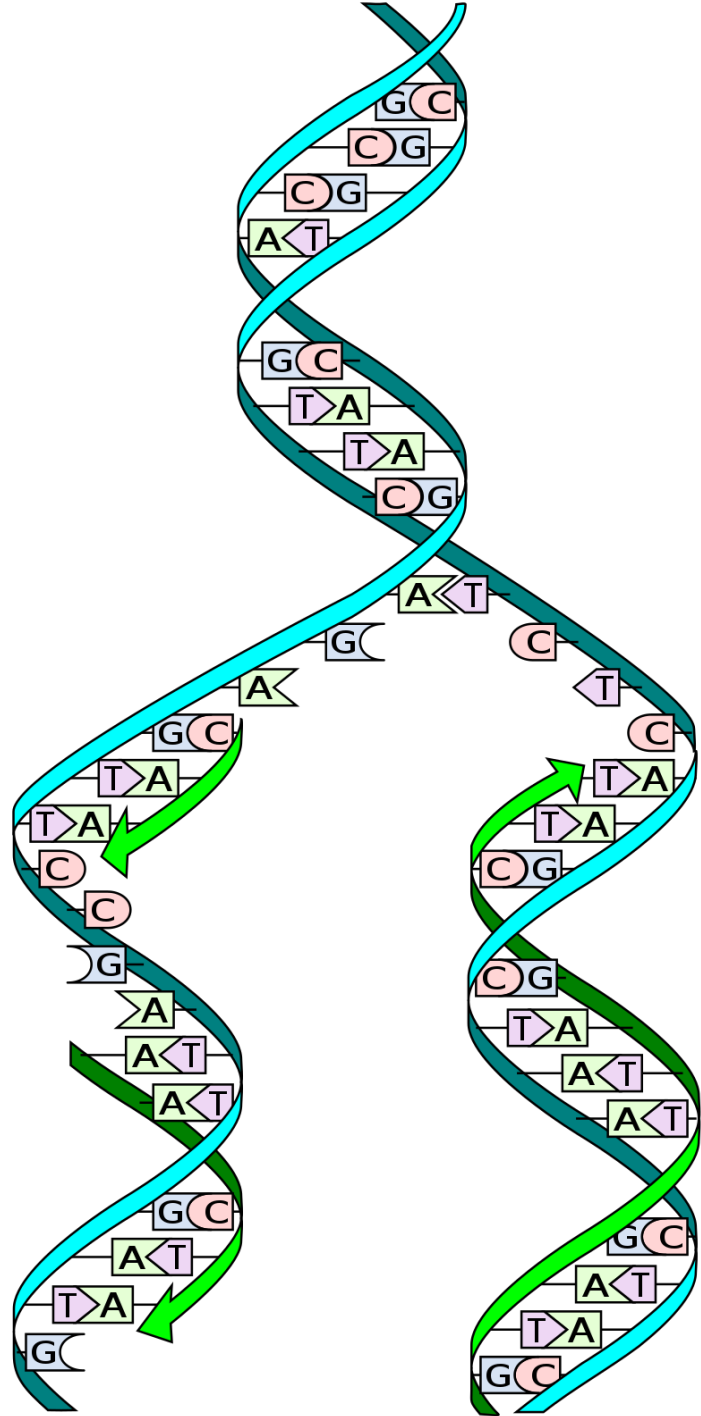
James Watson

Frances Crick

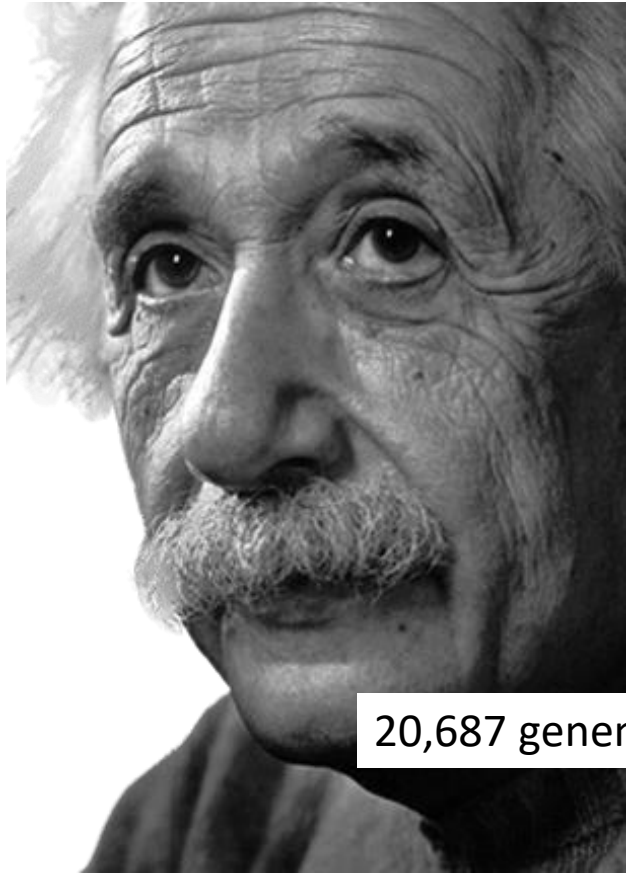
Rosalind Franklin



replicatie







20,687 genen

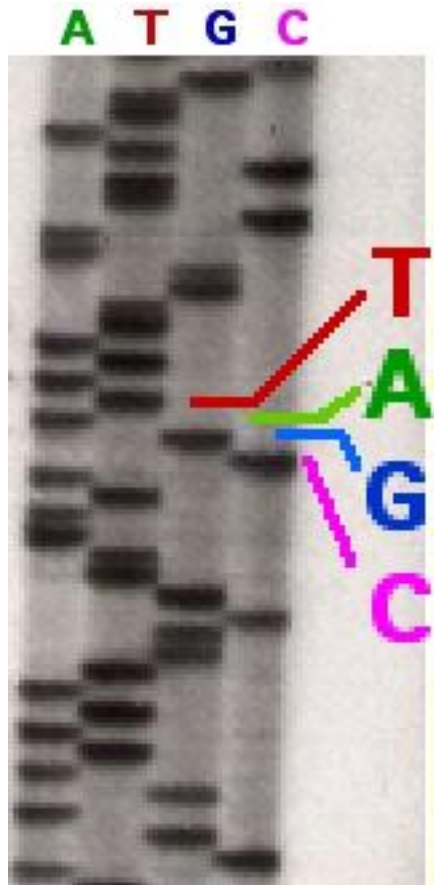
20,000 genen



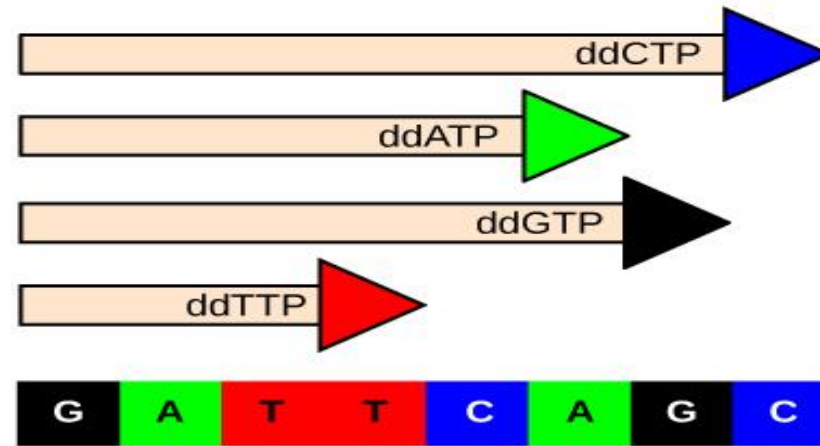
50,000 genen

Sanger sequencing

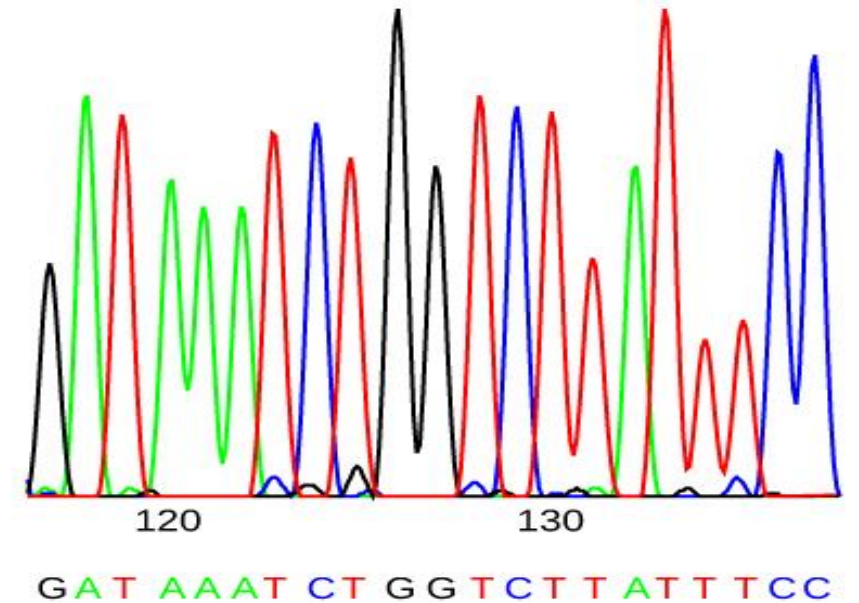
Ontwikkeld door Frederick Sanger et al. in 1977.



Incorporation fluorescent-labelled nucleotides which then stops the DNA replicating
Produces DNAs of different lengths ending with either A/T/G/C fluorescence



Dye-labeled dideoxynucleotides are used to generate DNA fragments of different lengths



Graph read out from fluorescent detector



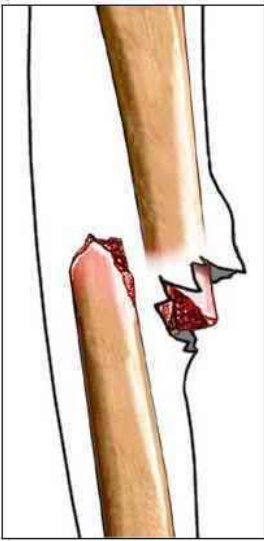
Whole exome
sequencing

Whole genome
sequencing



Osteogenesis Imperfecta (OI)

Blue sclerae

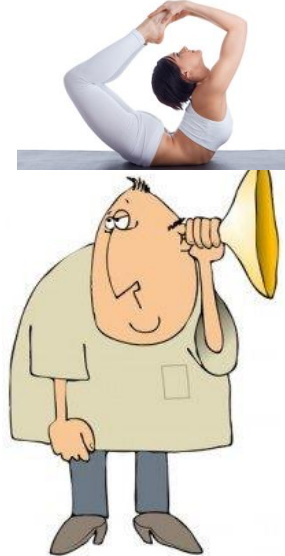


Fractures



Dentinogenesis Imperfecta

hypermobility

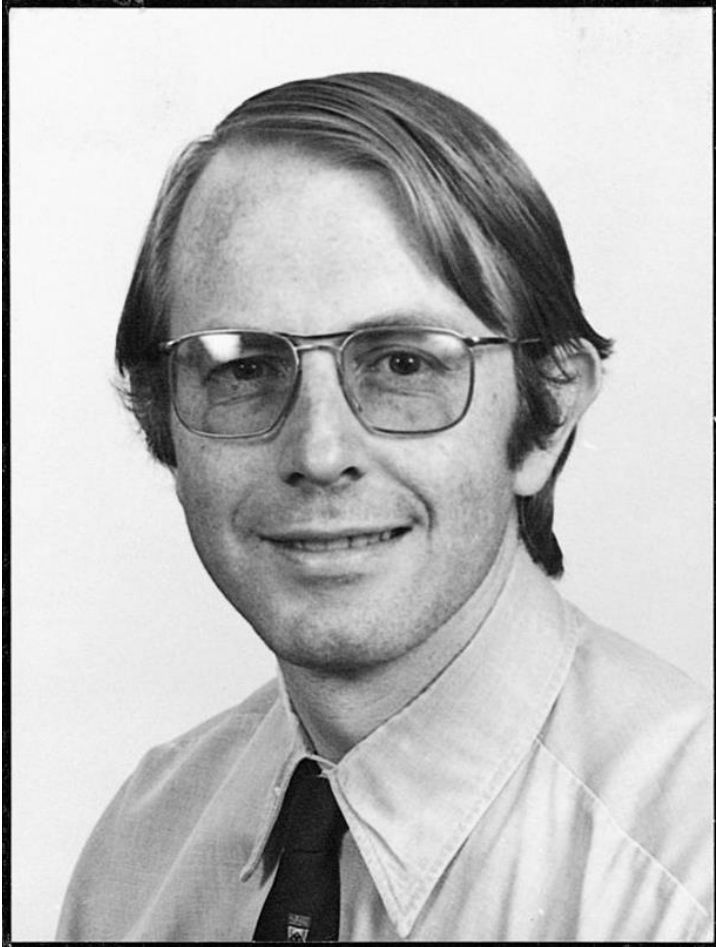


Hearing loss



Short stature

Prevalence 6-7/100,000



David Sillence schreef een artikel in 1979 waarin hij voorstelde om Ol te classificeren in 4 types n.a.v. klinische kenmerken, radiologie en overerving

Calcif. Tiss. Res. 17, 75—79 (1974)
© by Springer-Verlag 1974

Bone Collagen Aggregation Abnormalities in Osteogenesis Imperfecta

Steven L. Teitelbaum, William J. Kraft,
Robert Lang and Louis V. Avioli

Department of Pathology and Laboratory Medicine
Department of Medicine, Division of Endocrinology, The Jewish Hospital of St. Louis,
Washington University School of Medicine

Received May 17, accepted July 14, 1974

Scanning electron microscopy of bone collagen of three patients with osteogenesis imperfecta congenita demonstrates the failure of thin collagen fibers to aggregate extracellularly into the distinct and separately discernible large collagen fiber bundles of normal bone. These organizational alterations may in part explain the changes in physical properties which result in the frequent fractures characterizing this disease.

Key words: Bone — Collagen — Osteogenesis Imperfecta.

“Unraveling collagen” by Julian Voss-Andreae

Orange Memorial Park Sculpture Garden, City of South San Francisco, CA

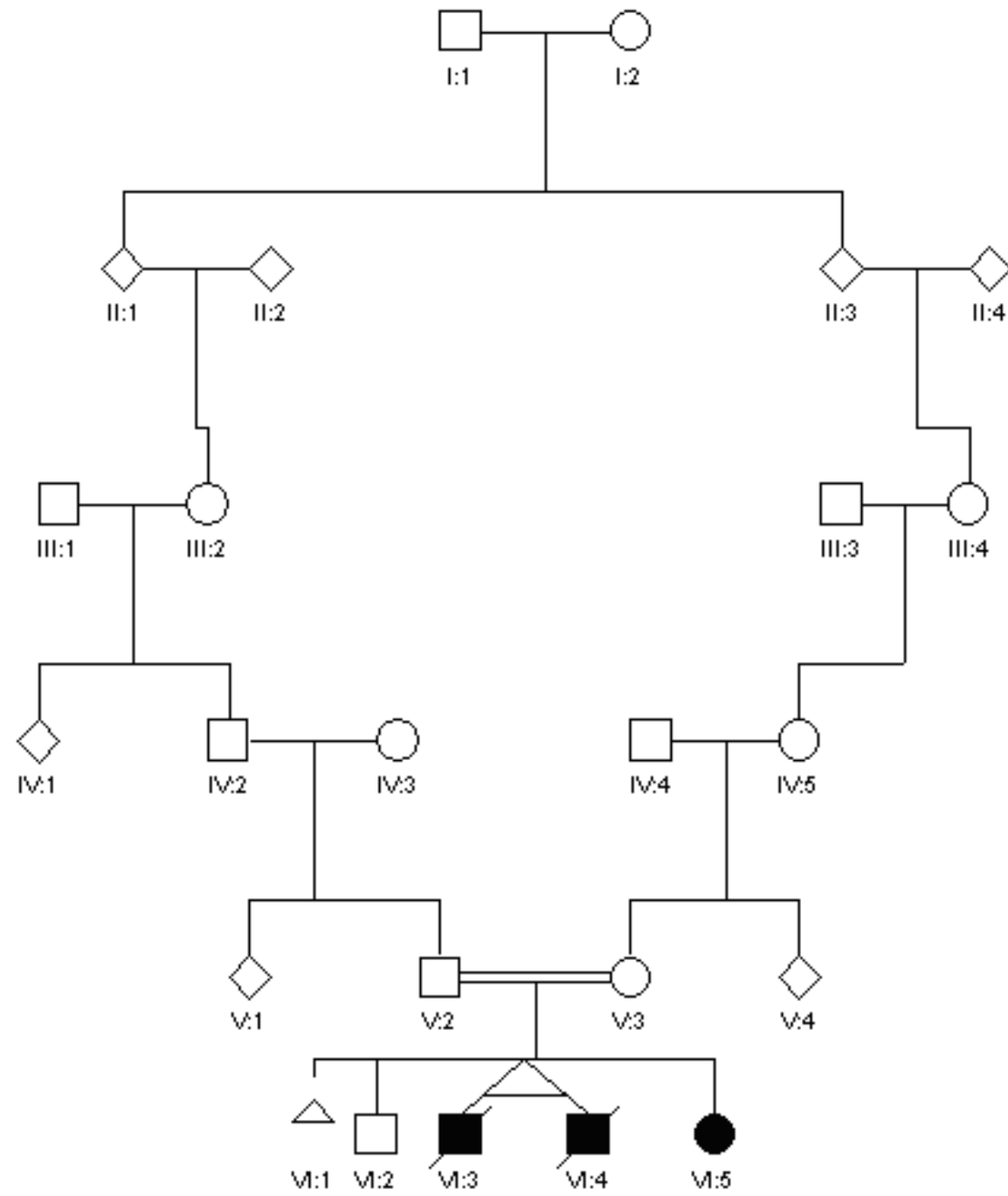


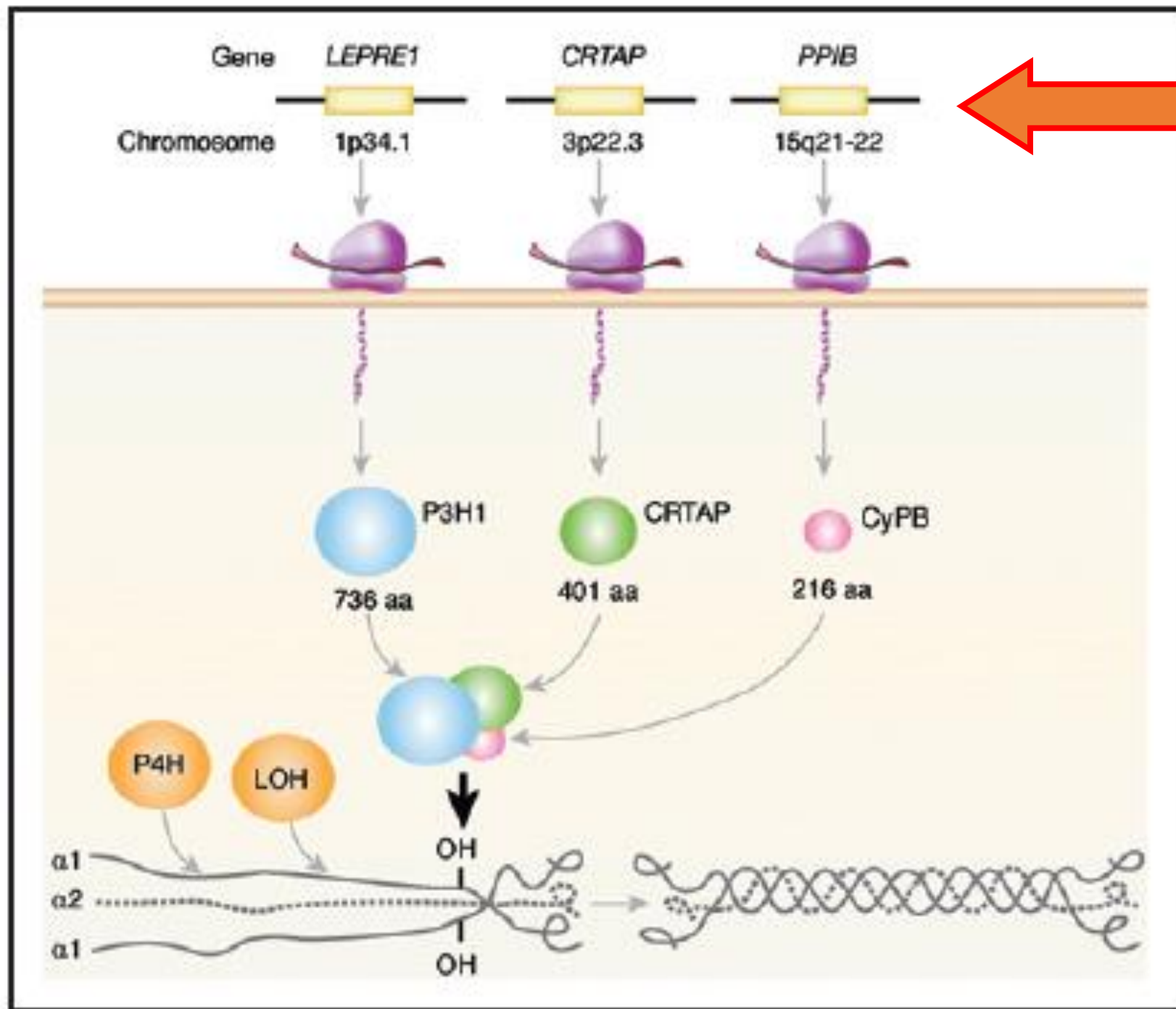
COL1A1

COL1A2

agtctatgctaattggccaattgcagcattaa

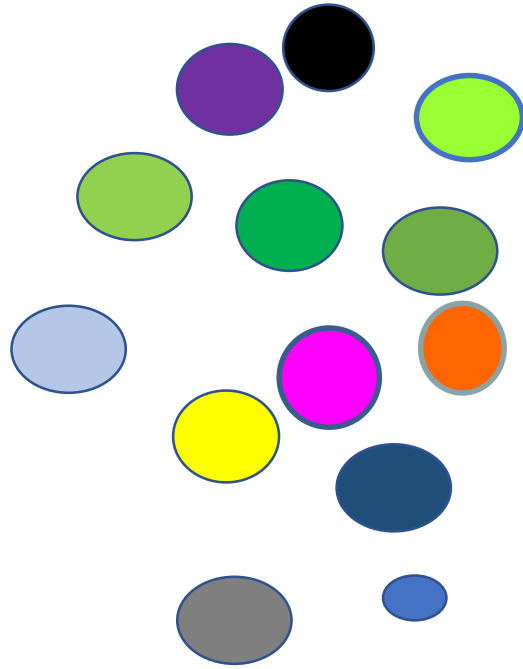
Baby met OI





welkom op de wereld





1



2



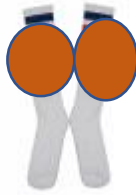
3



4



5



6



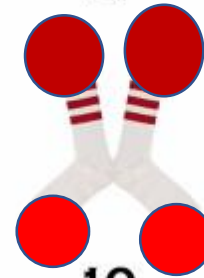
7



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22

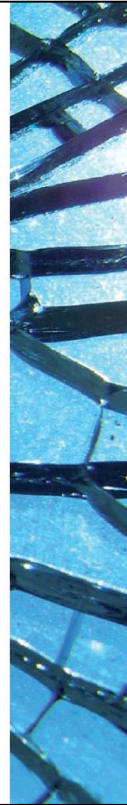


X Y



Thesis defense 16-12-11

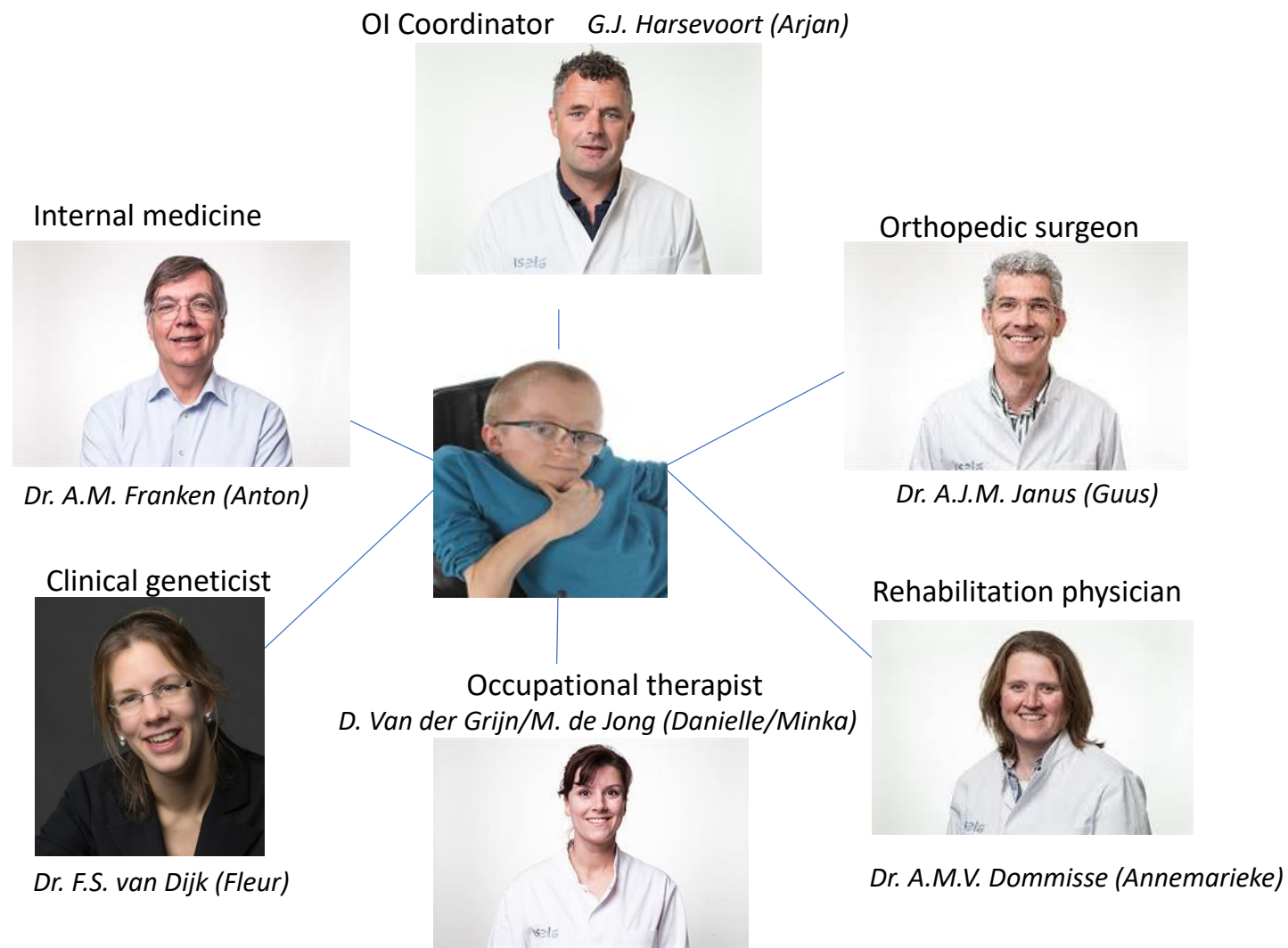
Osteogenesis Imperfecta;
Clinical and Genetic
Heterogeneity



Fleur van Dijk

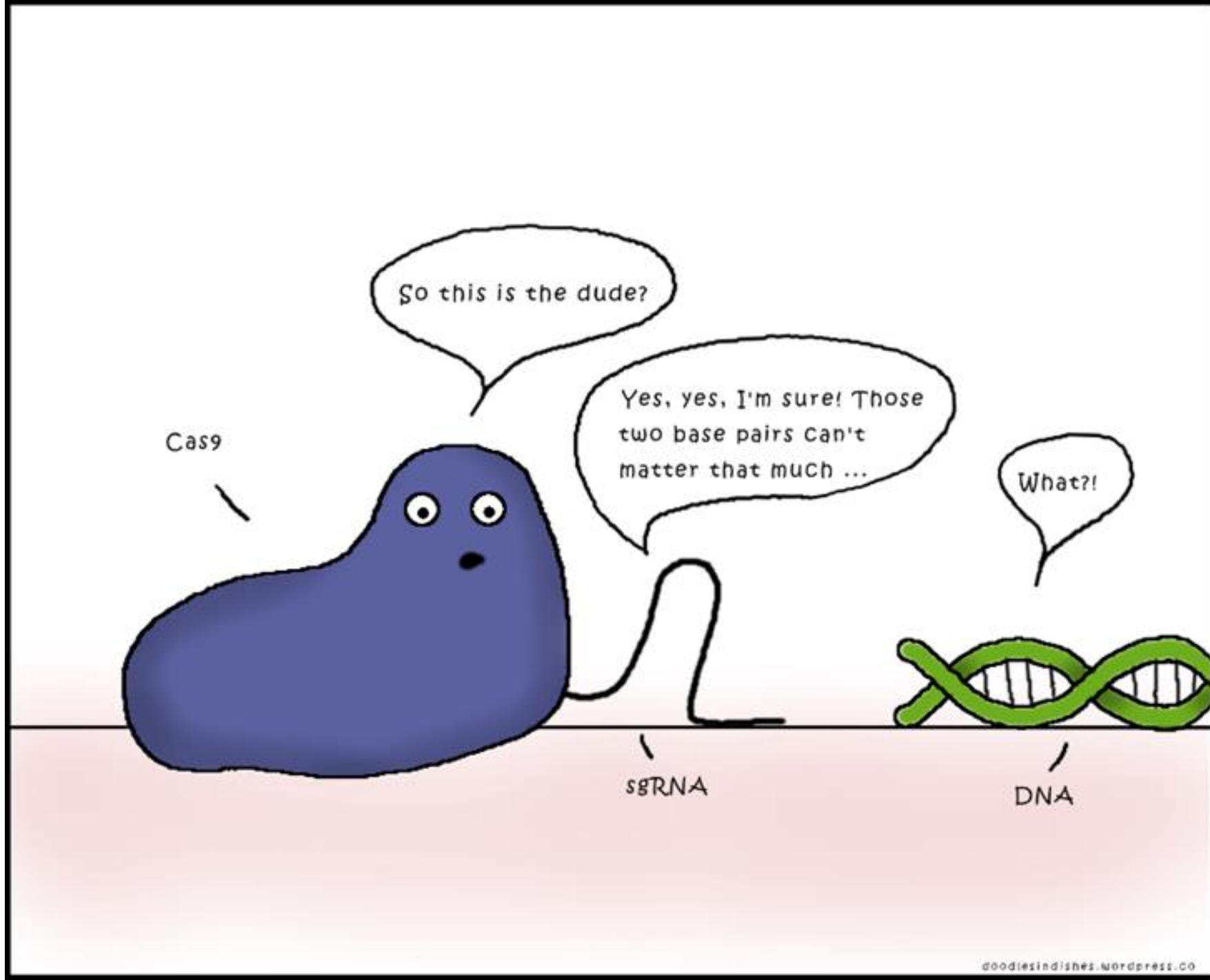






Ultimate cure







Use of induced pluripotent stem cells and CRISPR/Cas9 in Osteogenesis Imperfecta:

First results

Fleur S. van Dijk



LONDON: 2016



Expert center for complex Ehlers-Danlos Syndromes (EDS)

- London (South of UK) and Sheffield (North of UK)
- Diagnostic service for monogenic EDS subtypes



National Health Service: 70 years

- The NHS was created on July 5 1948 out of the ideal that good healthcare should be available to all, regardless of wealth.
- Geen verplichte zorgverzekering
- Gefinancierd door belasting en overheidsinkomsten

- Zorgsysteem zwaar overbelast
- Lange wachttijden
- Veel gecancelde operaties
- Tekort aan ambulances
- Private health sector



Ziekenhuis in Londen



Separated by a Common Translation? How the British and the Dutch Communicate

Bart Rottier, MD,^{1*} Nannette Ripmeester, MA,² and Andrew Bush, MD³

Summary. The British and Dutch share a long naval-, war- and medical history, in good times as well as bad. Their language has a common Germanic origin, but the English people may use special ways to express values or opinions, from which the sometimes paradoxical meaning is not always clear to the other party. Particularly with the Dutch, renowned for their directness, this may cause confusion. We provide a comprehensive set of expressions, each with paired interpretations, to foster Anglo–Dutch cooperation. **Pediatr Pulmonol.** 2011; 46:409–411. © 2010 Wiley-Liss, Inc.

Key words: communication; review; cooperation.

Funding source: none reported.

TABLE 1—Translation Guide Social

What the British say	What the British mean	What the Dutch understand
I hear what you say	I disagree completely	He ^a accepts my point of view
You must come by for dinner sometime	Just being polite; Goodbye!	He will invite me for dinner in the course of time
Very interesting	I don't agree	He likes my idea
With the greatest respect	You must be a fool	He respects me/my view
I'm sure it's my fault	It's your fault!	It is his fault
That is an original point of view	You must be crazy	They like the idea
I almost agree	I don't agree	He almost agrees
You'll get there (eventually)	No way you will make it	Encouragement to go on
I'll bear it in mind	I won't do anything about it	He will use it when appropriate
Could we consider some other options	I don't like your idea	He is still in the process of thinking
I would suggest	Do it as I want you to	An open suggestion
By the way	The primary purpose is	Not very important
Perhaps you could give this some more thought	Don't do it, it's a bad idea	Consider possible road blocks
Quite good	A bit disappointing	Quite good
Not bad	(very) good	Average or poor

^aWhere “he” is mentioned “she” can also be read.