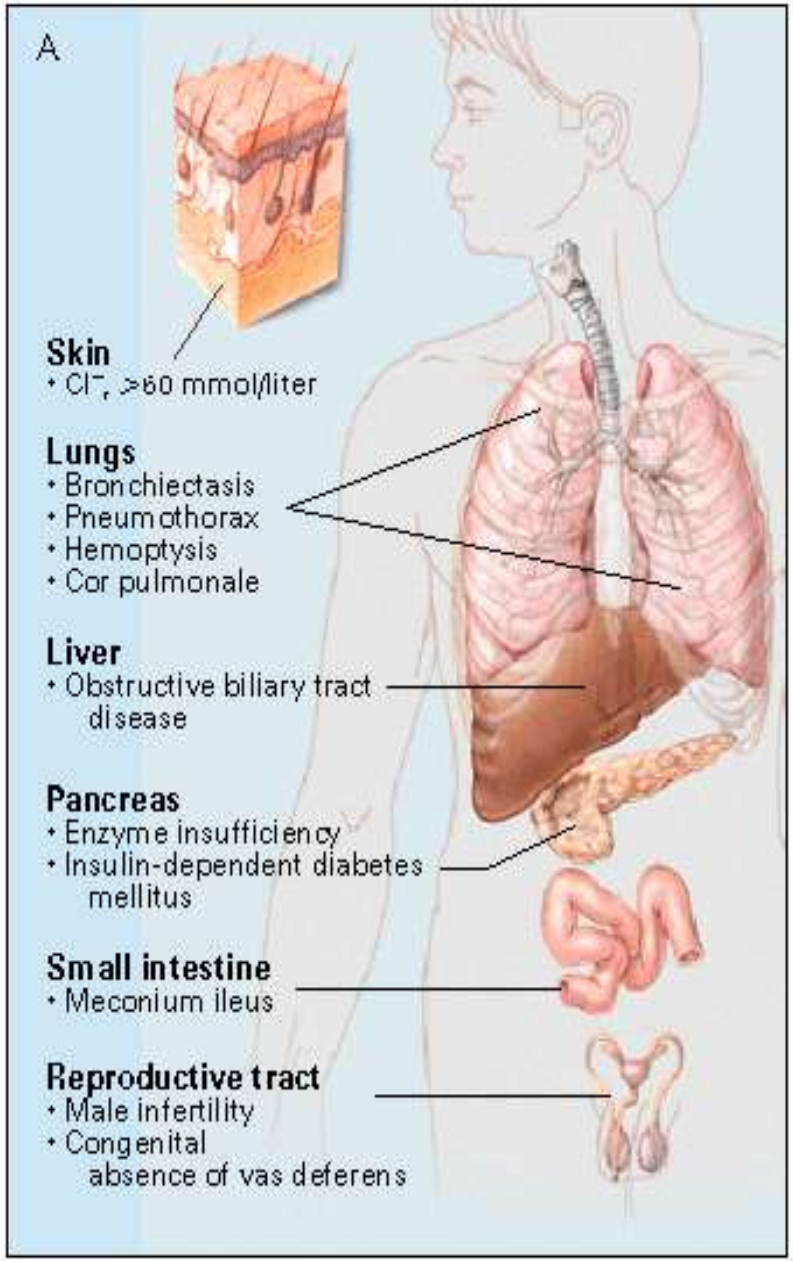
Perdiktivní genetika a její klinická užitečnost: na příkladu cystické fibrózy

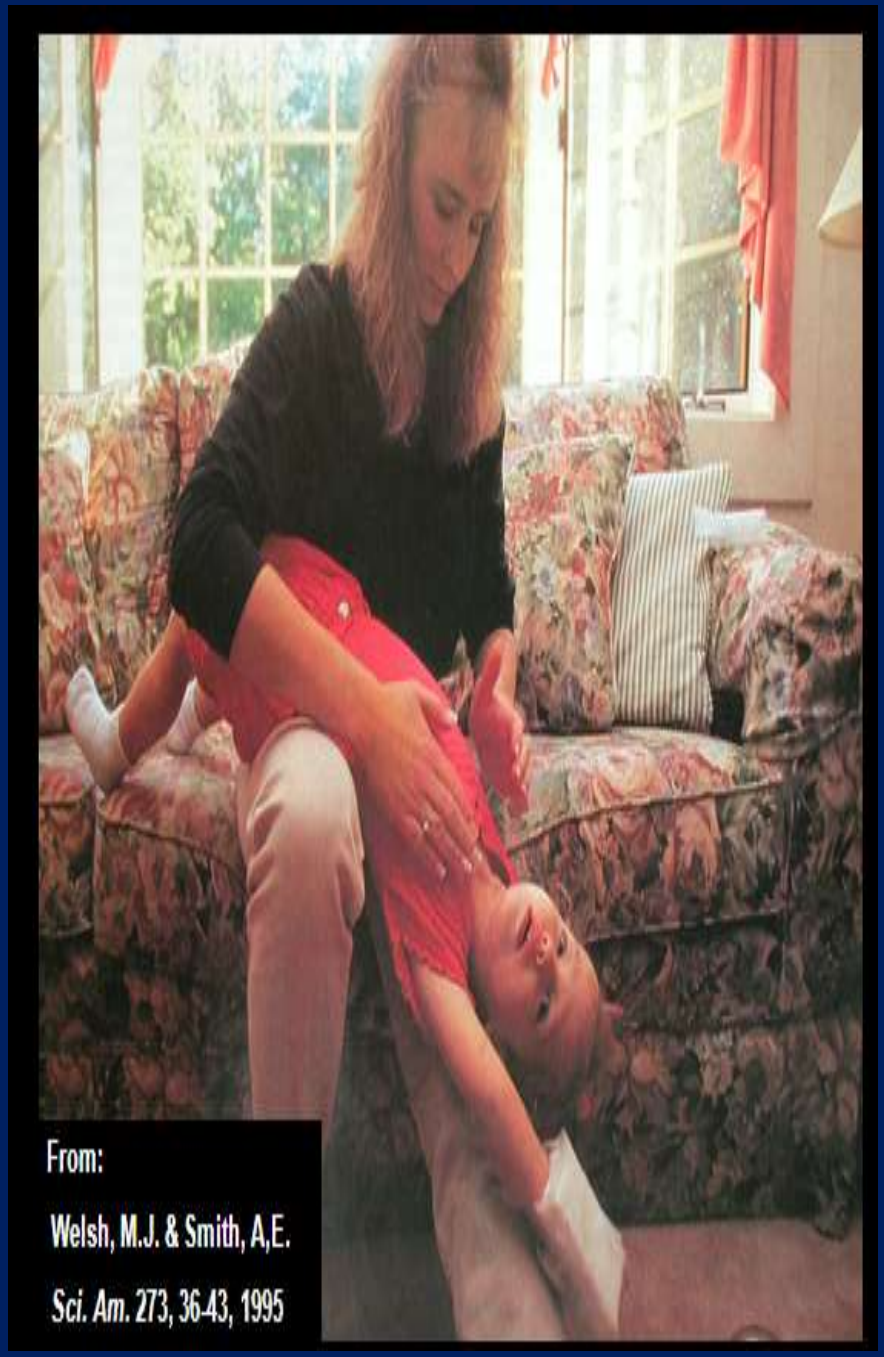
Workshop on Atopic Dermatitis
23-24. května 2008
Praha – Štefánikova Hvězdárna

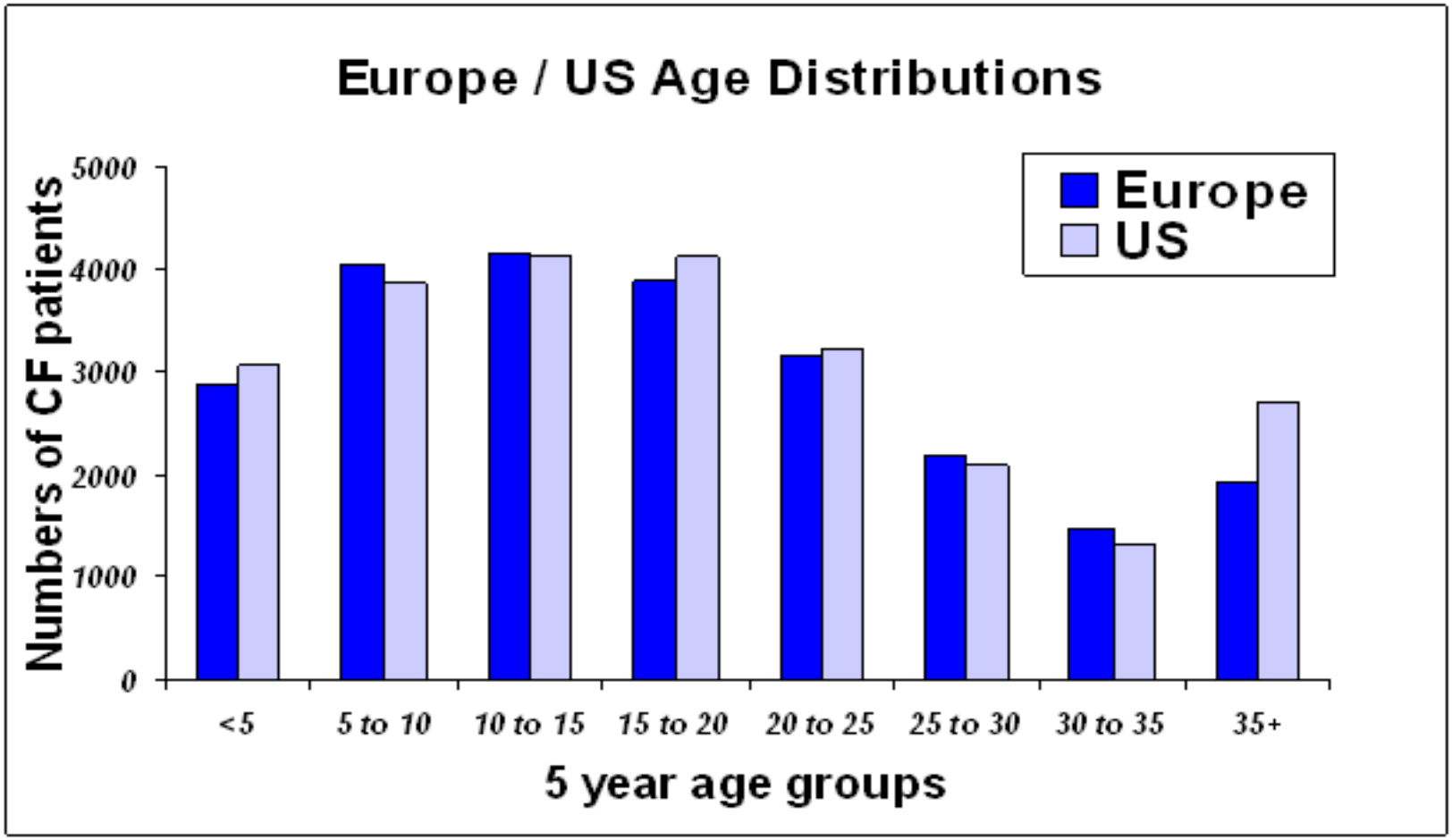
Milan Macek Jr.

Ústav biologie a lékařské genetiky – UK 2. LF a FN v Motole

ublg.lf2.cuni.cz


A
Skin
• Cl⁻, >60 mmol/liter
Lungs
• Bronchiectasis
• Pneumothorax
• Hemoptysis
• Cor pulmonale
Liver
• Obstructive biliary tract disease
Pancreas
• Enzyme insufficiency
• Insulin-dependent diabetes mellitus
Small intestine
• Meconium ileus
Reproductive tract
• Male infertility
• Congenital absence of vas deferens


From:
Welsh, M.J. & Smith, A.E.
Sci. Am. 273, 36-43, 1995



Patients from Europe: 23693, US: 24487, Total: 48180

21 countries from Europe + US data shown

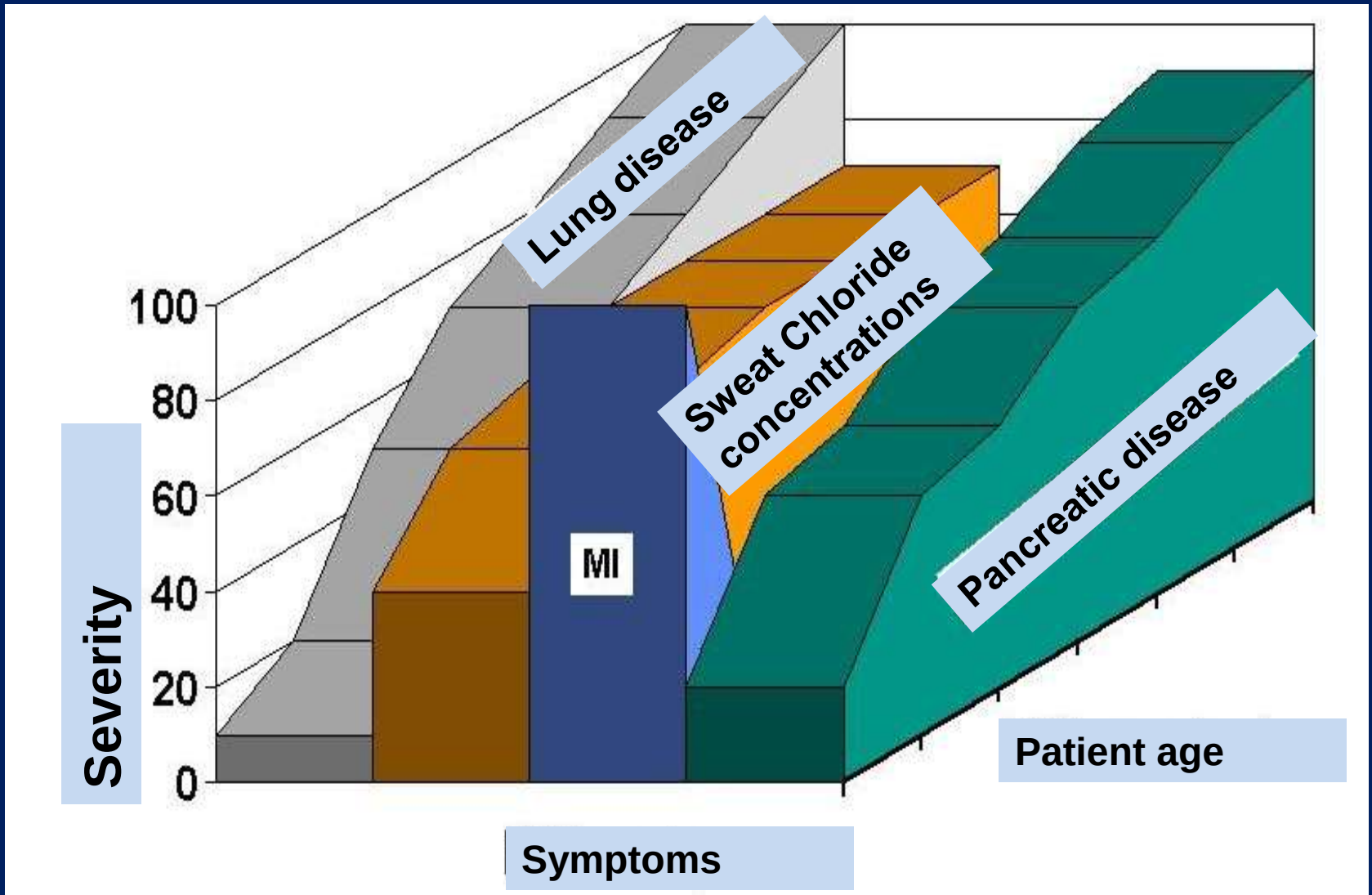
Data from CFF Registry, ECFS & EuroCareCF

UNIVERSITY OF DUNDEE

EuroCareCF

wwww.eurocarecf.eu, www.ecfs.eu, www.cff.org

CF is a syndrome



Medieval Nursery Rhymes: empirical evidence of CF

Wehe dem kind
Das beim kuss auf die Stirn
Salzig schmeckt
Es ist Överhext und muss bald sterben

Die Mutter küsst das Kind
Es schmeckt salzig
Sie weiss Bescheid
Es wird nicht alt

„Kiss your baby“

- A few references uncovered in medieval folklore predict death for an infant that tastes "salty" when kissed. Such infants were thought to be "hexed“

- In 1606, Alonso y de los Ruyzes de Fonteca, professor of medicine at Henares in Spain, wrote that it was known that the fingers taste salty after rubbing the forehead of the bewitched child

1810-
1839

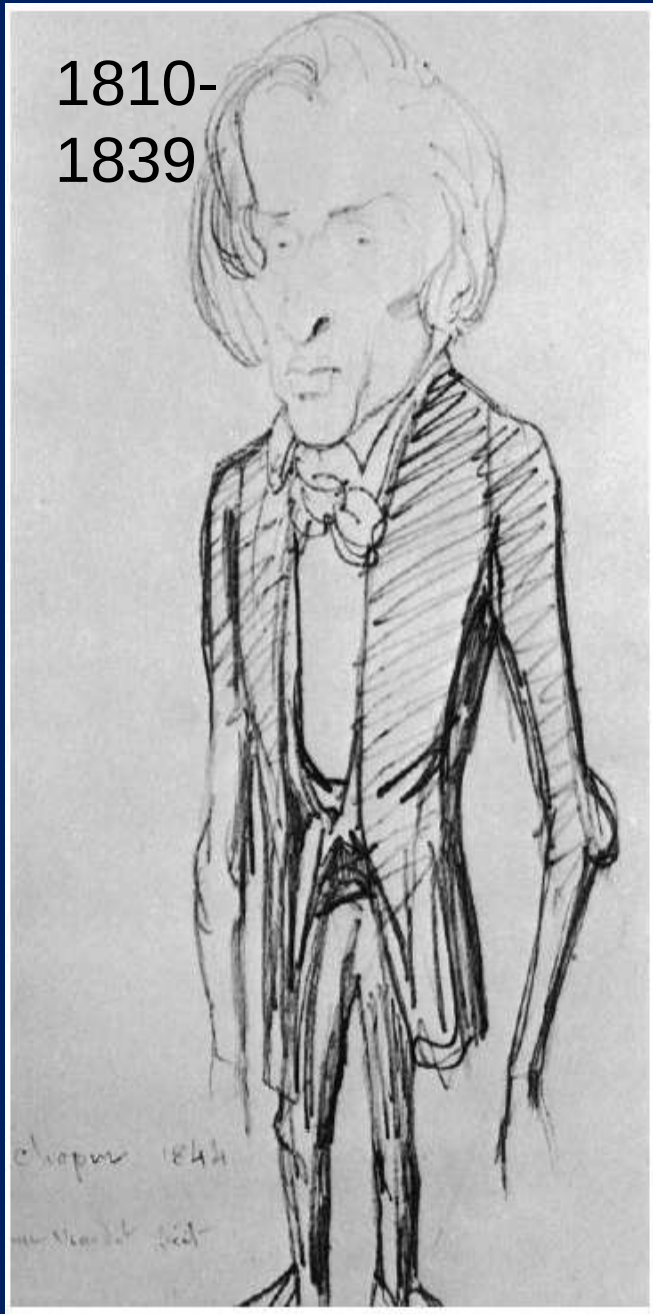


FIGURE 2. A caricature done in 1844 shows a barrel-chested Chopin with thin limbs (courtesy of Madam M. Maurvois).

(1) Recurrent respiratory complaints (coughing, breathlessness, hemoptysis,respiratoryInsufficiency) which seem to have started in the patient's teens;

(2) Systemic complaints (poor exercise tolerance, tiredness, emaciation, failure to gain weight, pallor, pigmentation, peripheral edema, muscle wasting, and icterus);

(3) GI symptoms (diarrhea, fatty food intolerance, hematemesis)

(4) Infertility

(4) no finger clubbing

Majka et al. Cystic fibrosis--a probable cause of Frederic Chopin's suffering and death. J Appl Genet. 2003;44(1):77-84.
Kubba et al. The long suffering of Frederic Chopin. Chest. 1998 Jan;113(1):210-6.

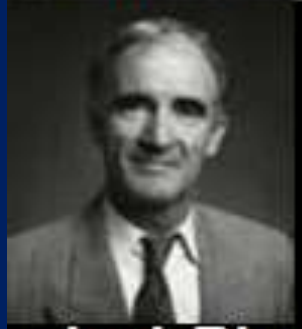
Dorothy Andersen 1938

CYSTIC FIBROSIS

(Cystic Fibrosis of the Pancreas or Mucoviscidosis)

A complex genetic disease affecting a number of organs systems including the lung and upper respiratory tract, the gastrointestinal tract, pancreas, liver, sweat glands and the genitourinary tract.

Francis Collins

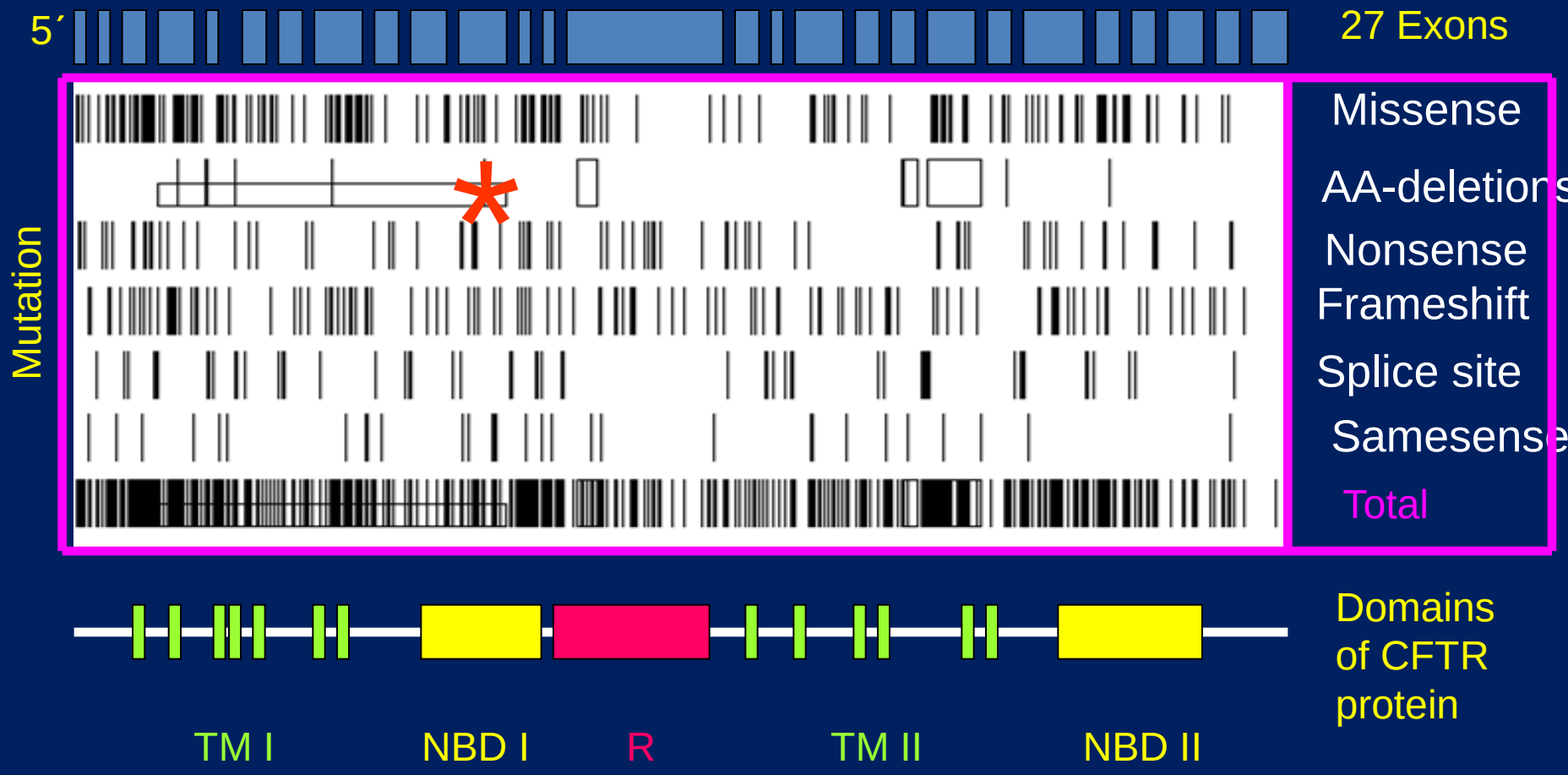


Jack Riordan

Lap-Chee Tsui

8th September 1989

CFTR gene *(MIM: 602421)*



Mean frequency of F508del mutation in European-derived populations ~ 70%.
Since 1989 over 1 500 germinal CFTR mutations have been identified-

www.genet.sickkids.on.ca/cftr

Motto

Stars and Mutations

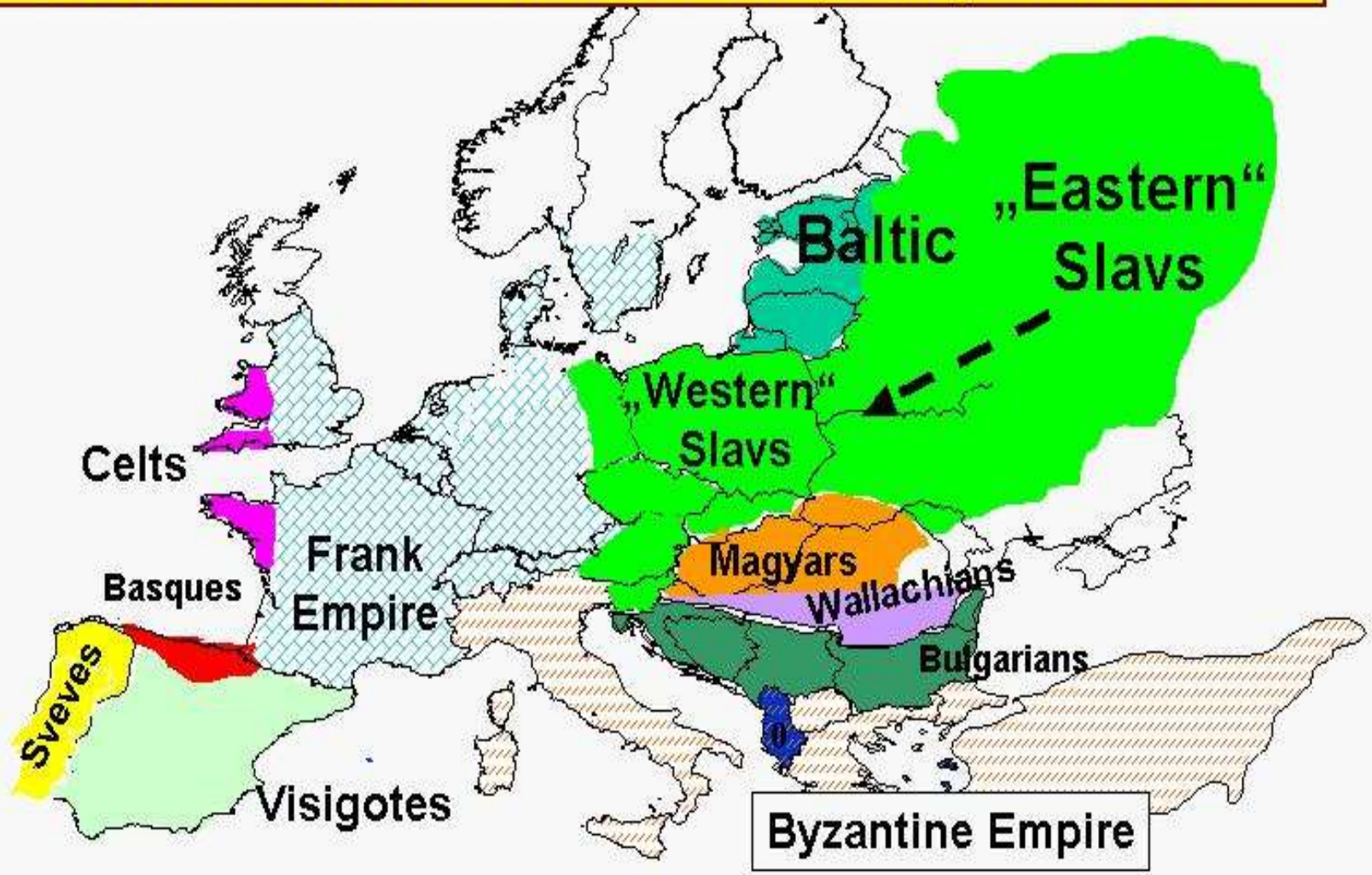
Mutations are similar to the light of particular stars that we see on the bright night sky. Since their light has travelled to us over several million (- billion) light-years, what we now mostly see in „real-time“ is a „message“ from already extinct stars.

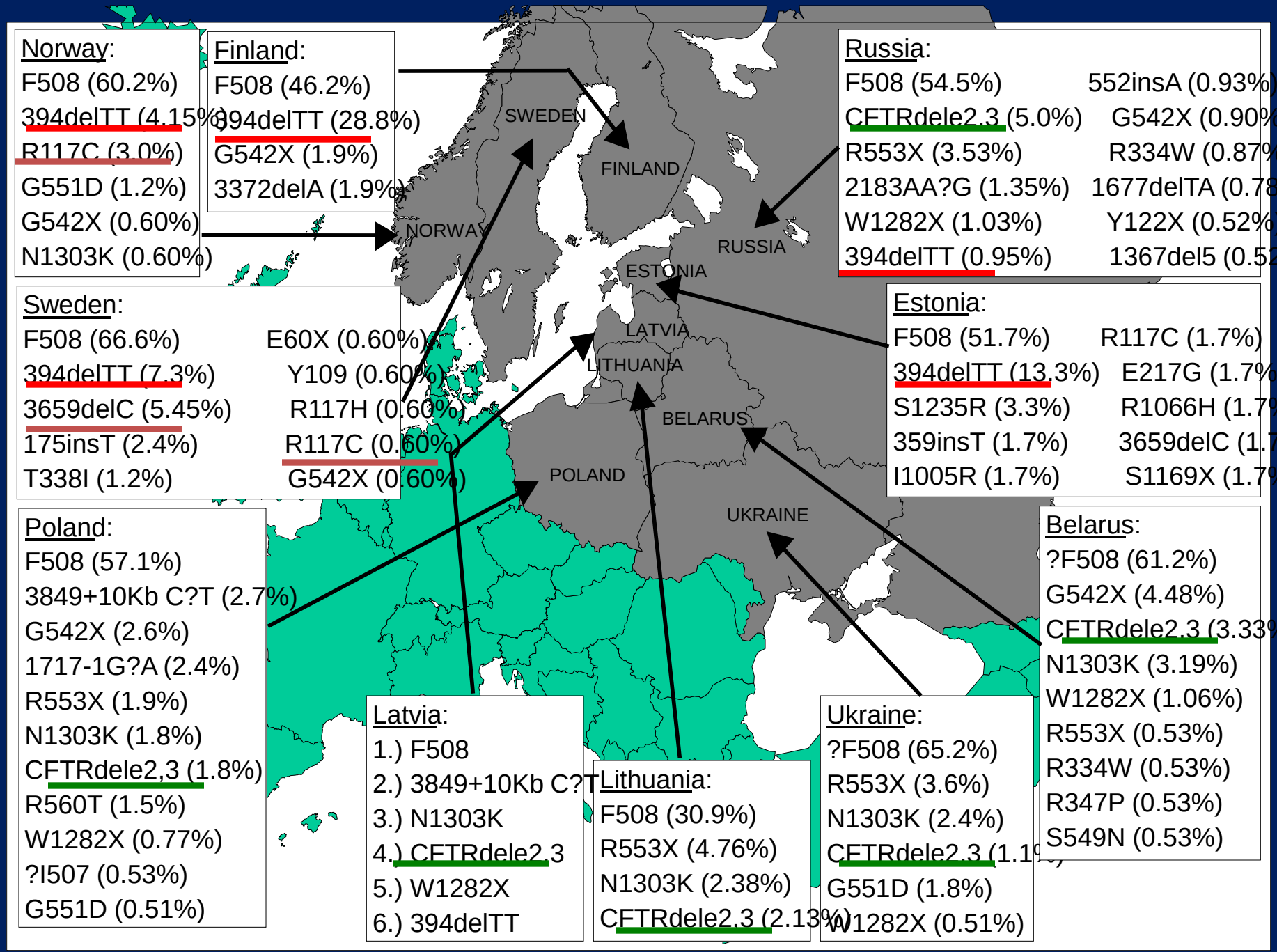
Thus, mutations convey a similar „message“ from already extinct ancestral populations, that have however significantly influenced our current gene pool.

Prof. A. Piazza, Italy

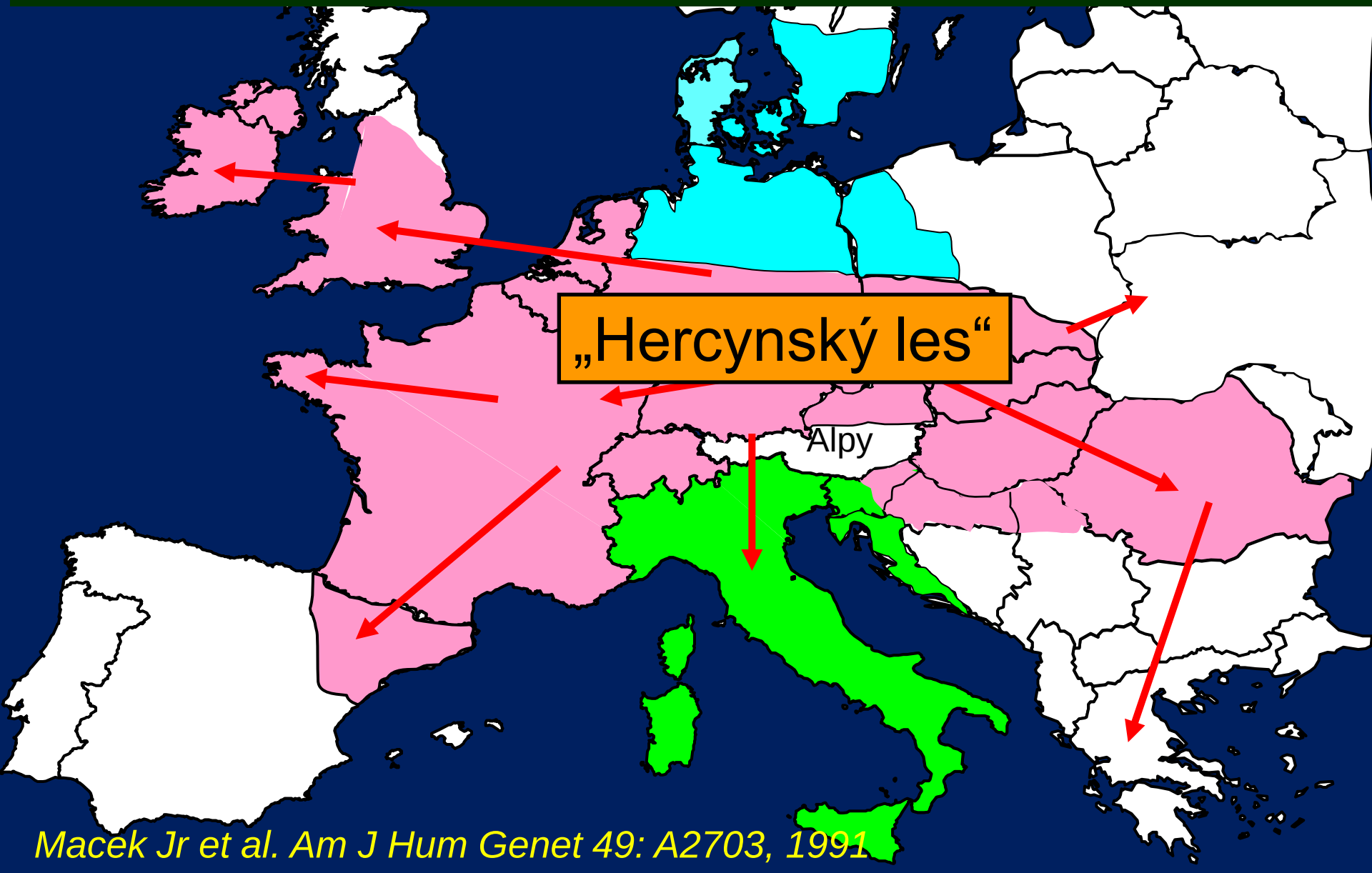
Cohorts > 1000 CF chromosomes

European populations at the 5th C.A.D. and the distribution of *CFTR*dele2,3(21kb) mutation



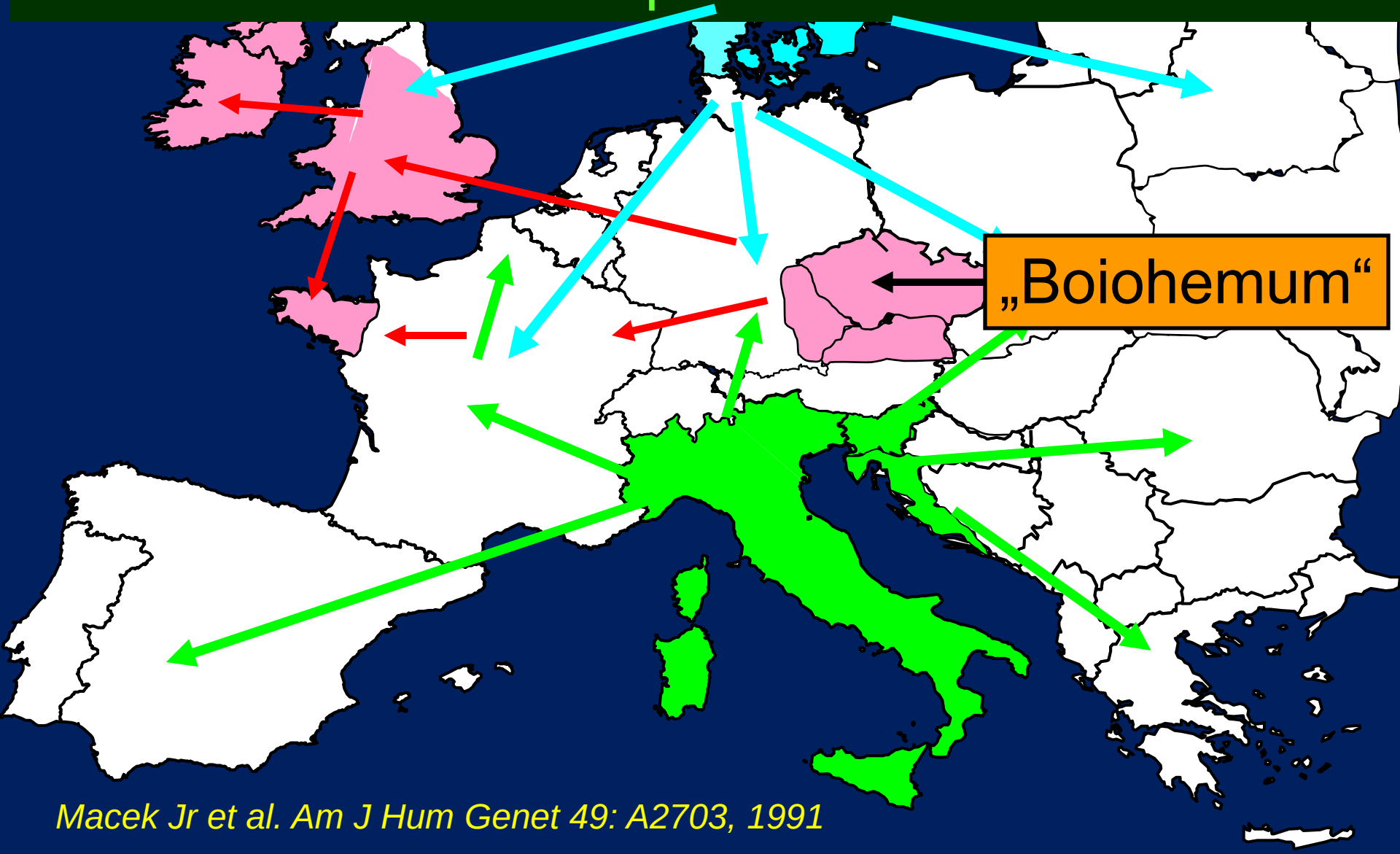

Norway:
F508 (60.2%)
394delTT (4.15%)
R117C (3.0%)
G551D (1.2%)
G542X (0.60%)
N1303K (0.60%)
Finland:
F508 (46.2%)
394delTT (28.8%)
G542X (1.9%)
3372delA (1.9%)
Russia:
F508 (54.5%)
552insA (0.93%)
CFTRdele2,3 (5.0%)
G542X (0.90%)
R553X (3.53%)
R334W (0.87%)
2183AA?G (1.35%)
1677delTA (0.78%
W1282X (1.03%)
Y122X (0.52%)
394delTT (0.95%)
1367del5 (0.52%
Sweden:
F508 (66.6%)
E60X (0.60%)
394delTT (7.3%)
Y109 (0.60%)
3659delC (5.45%)
R117H (0.60%)
175insT (2.4%)
R117C (0.60%)
T338I (1.2%)
G542X (0.60%)
Estonia:
F508 (51.7%)
R117C (1.7%)
394delTT (13.3%)
E217G (1.7%)
S1235R (3.3%)
R1066H (1.7%)
359insT (1.7%)
3659delC (1.7%
I1005R (1.7%)
S1169X (1.7%)
Poland:
F508 (57.1%)
3849+10Kb C?T (2.7%)
G542X (2.6%)
1717-1G?A (2.4%)
R553X (1.9%)
N1303K (1.8%)
CFTRdele2,3 (1.8%)
R560T (1.5%)
W1282X (0.77%)
?I507 (0.53%)
G551D (0.51%)
Belarus:
?F508 (61.2%)
G542X (4.48%)
CFTRdele2,3 (3.33%)
N1303K (3.19%)
W1282X (1.06%)
R553X (0.53%)
R334W (0.53%)
R347P (0.53%)
S549N (0.53%)
Latvia:
1.) F508
2.) 3849+10Kb C?T
3.) N1303K
4.) CFTRdele2,3
5.) W1282X
6.) 394delTT
Lithuania:
F508 (30.9%)
R553X (4.76%)
N1303K (2.38%)
CFTRdele2,3 (2.13%)
Ukraine:
?F508 (65.2%)
R553X (3.6%)
N1303K (2.4%)
CFTRdele2,3 (1.1%)
G551D (1.8%)
W1282X (0.51%)
SWEDEN
FINLAND
NORWAY
RUSSIA
ESTONIA
LATVIA
LITHUANIA
BELARUS
POLAND
UKRAINE

Rozšíření keltské civilizace před expanzí Nordických kmenů a Římského Impéria



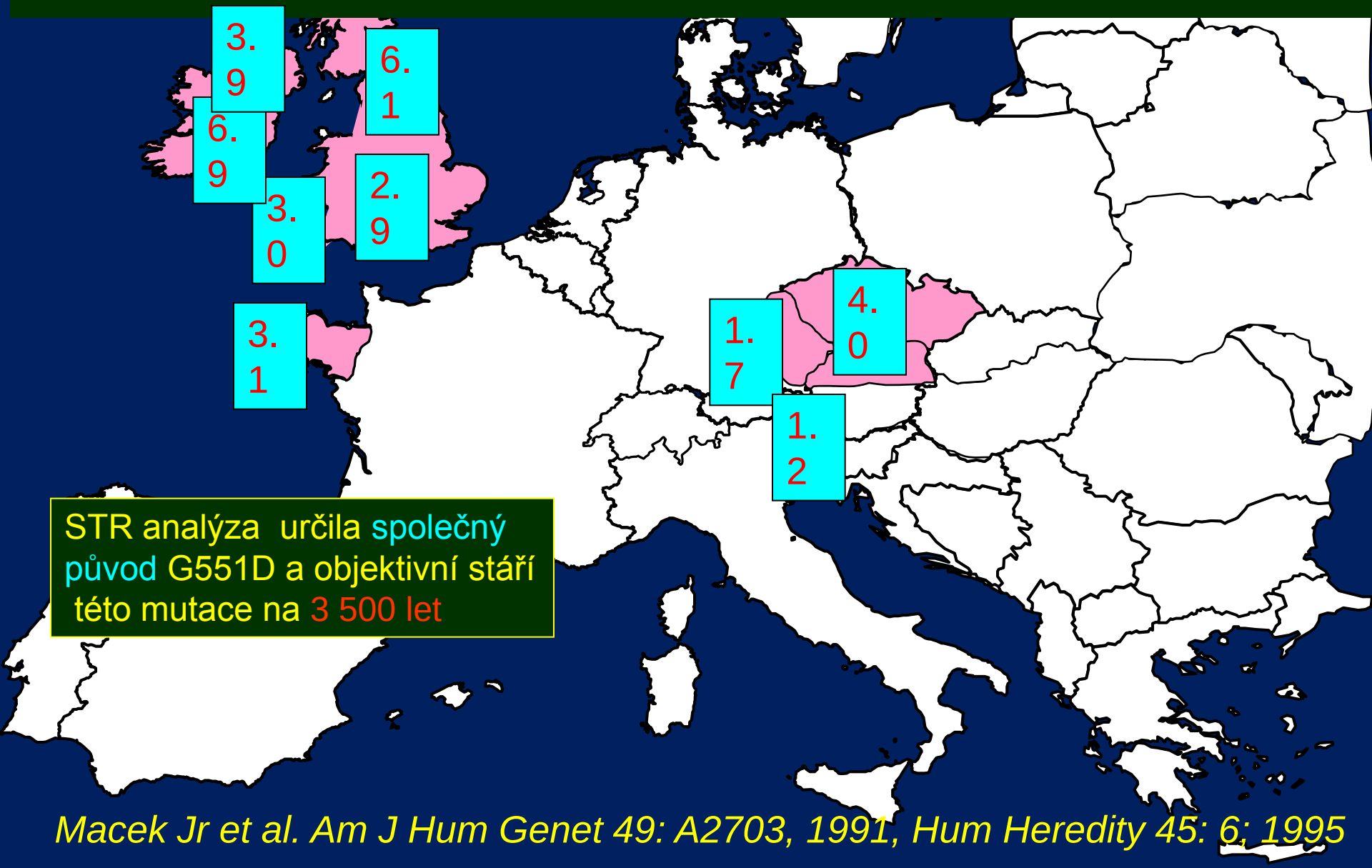
Macek Jr et al. Am J Hum Genet 49: A2703, 1991

Vyhnání keltských kmenů ze Střední a Západní Evropy Nordickými kmeny and Římským Impériem



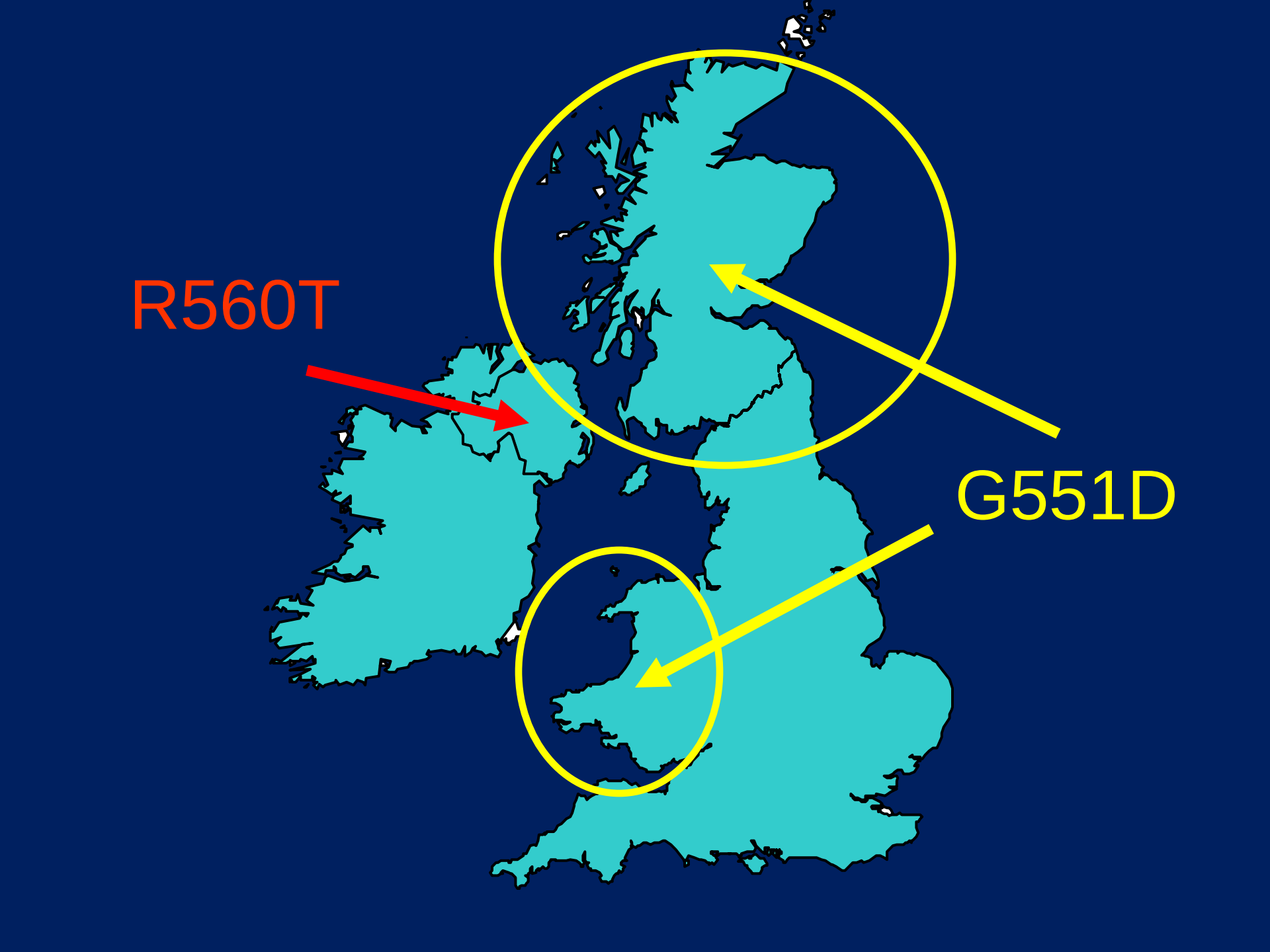
Macek Jr et al. Am J Hum Genet 49: A2703, 1991

Současný výskyt „keltské“ mutace G551D

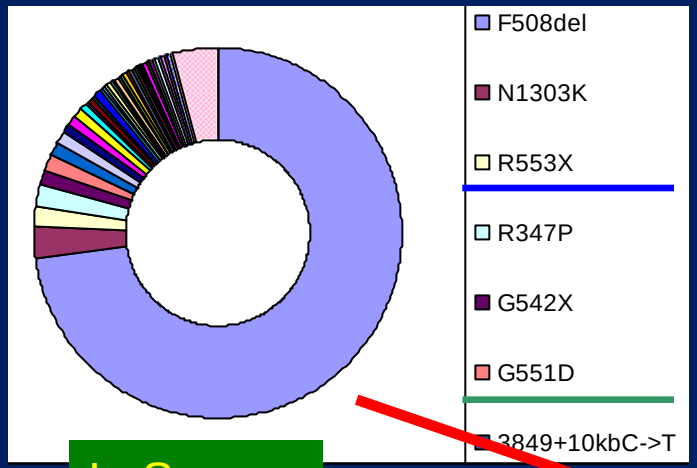


STR analýza určila společný původ G551D a objektivní stáří této mutace na 3 500 let

Macek Jr et al. Am J Hum Genet 49: A2703, 1991, Hum Heredity 45: 6; 1995

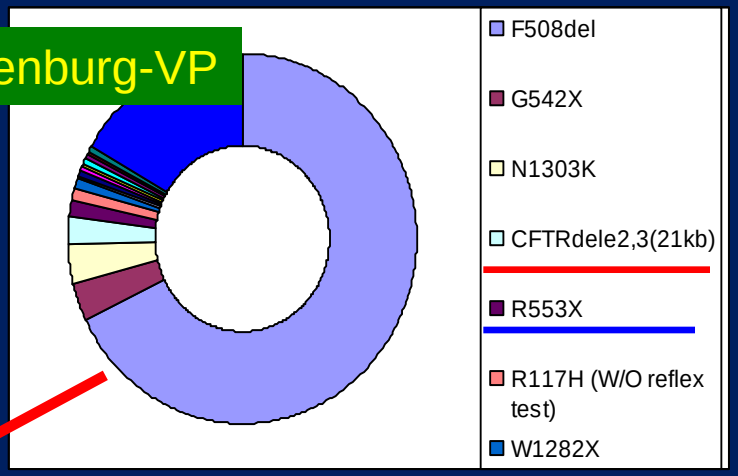

R560T
G551D

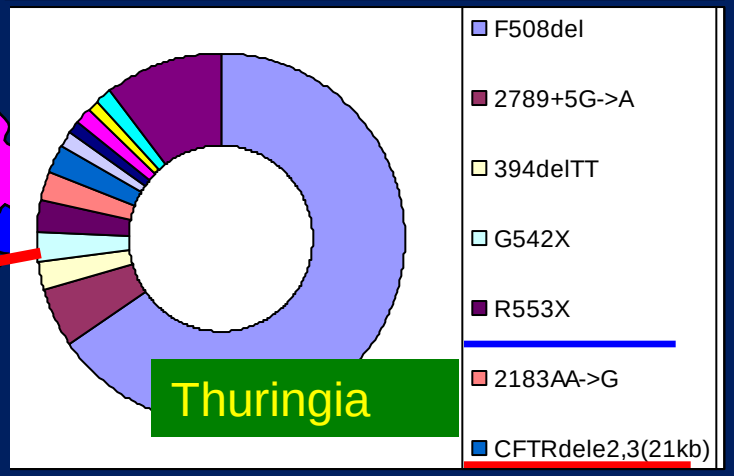
Germany



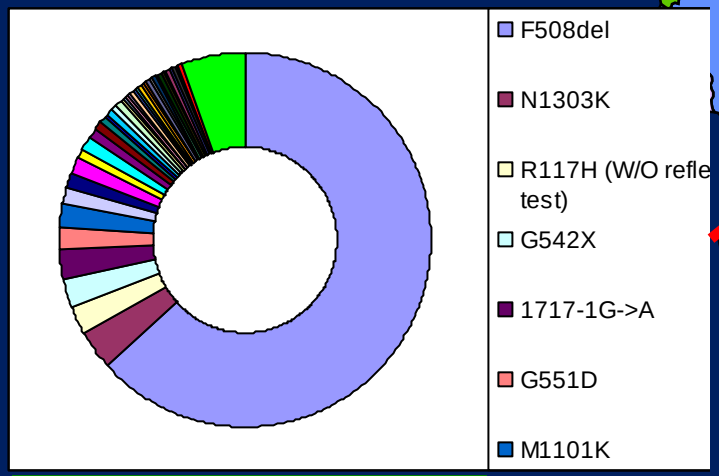
L. Saxony

Mecklenburg-VP

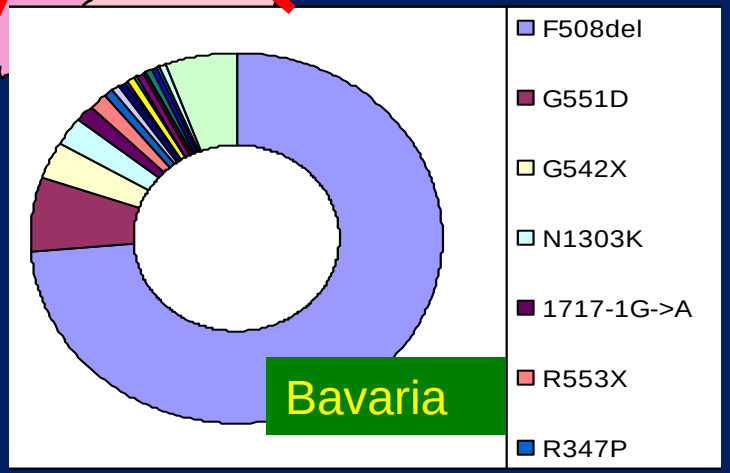




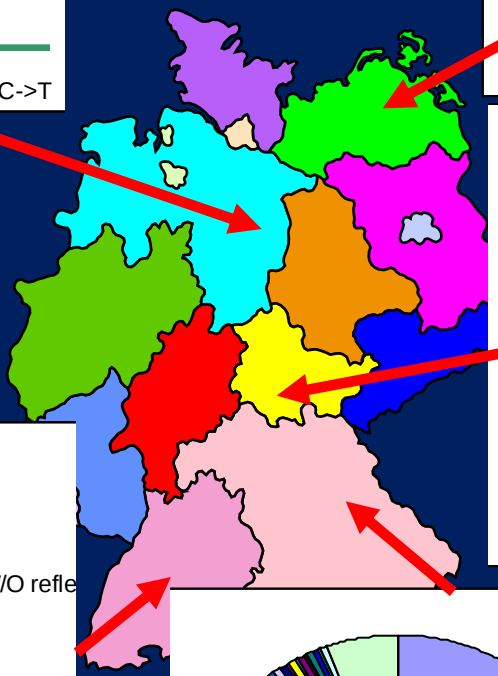
Thuringia



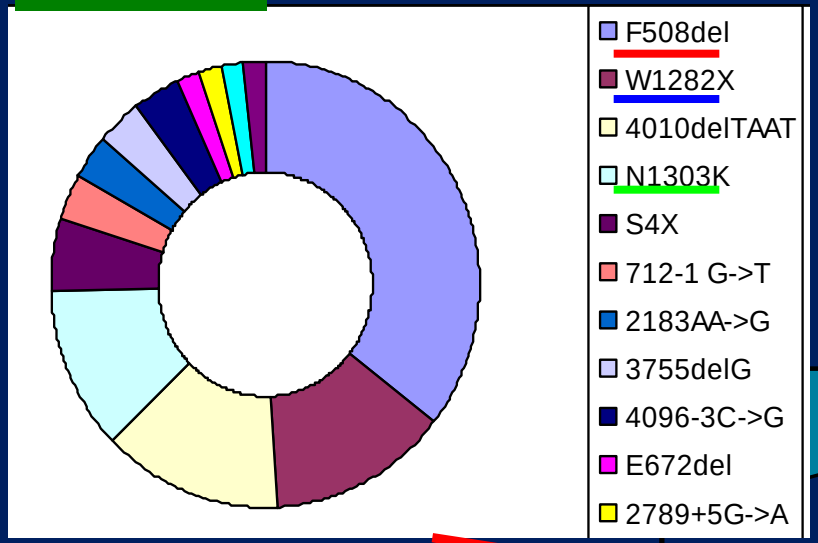
Baden-Wuertenberg



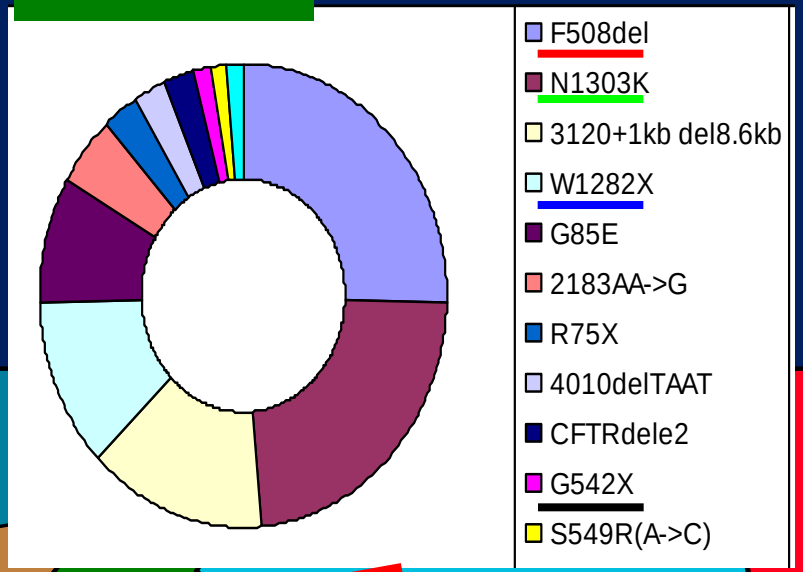
Bavaria



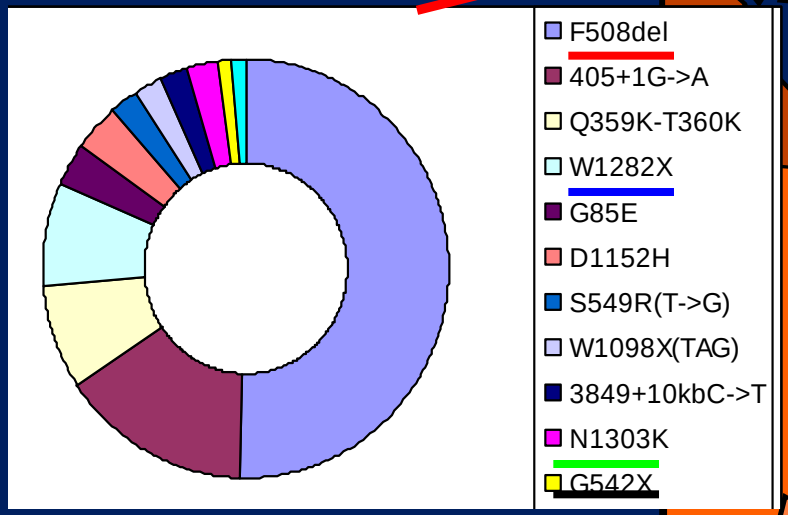
Lebanese



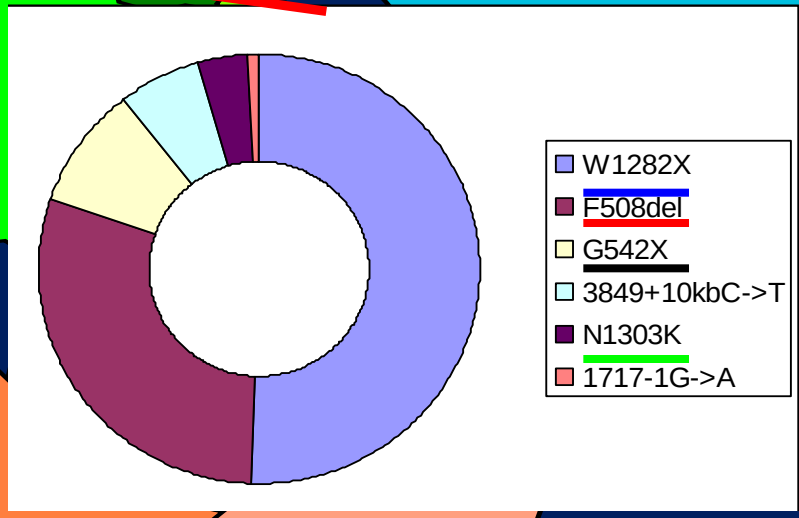
Palestinians



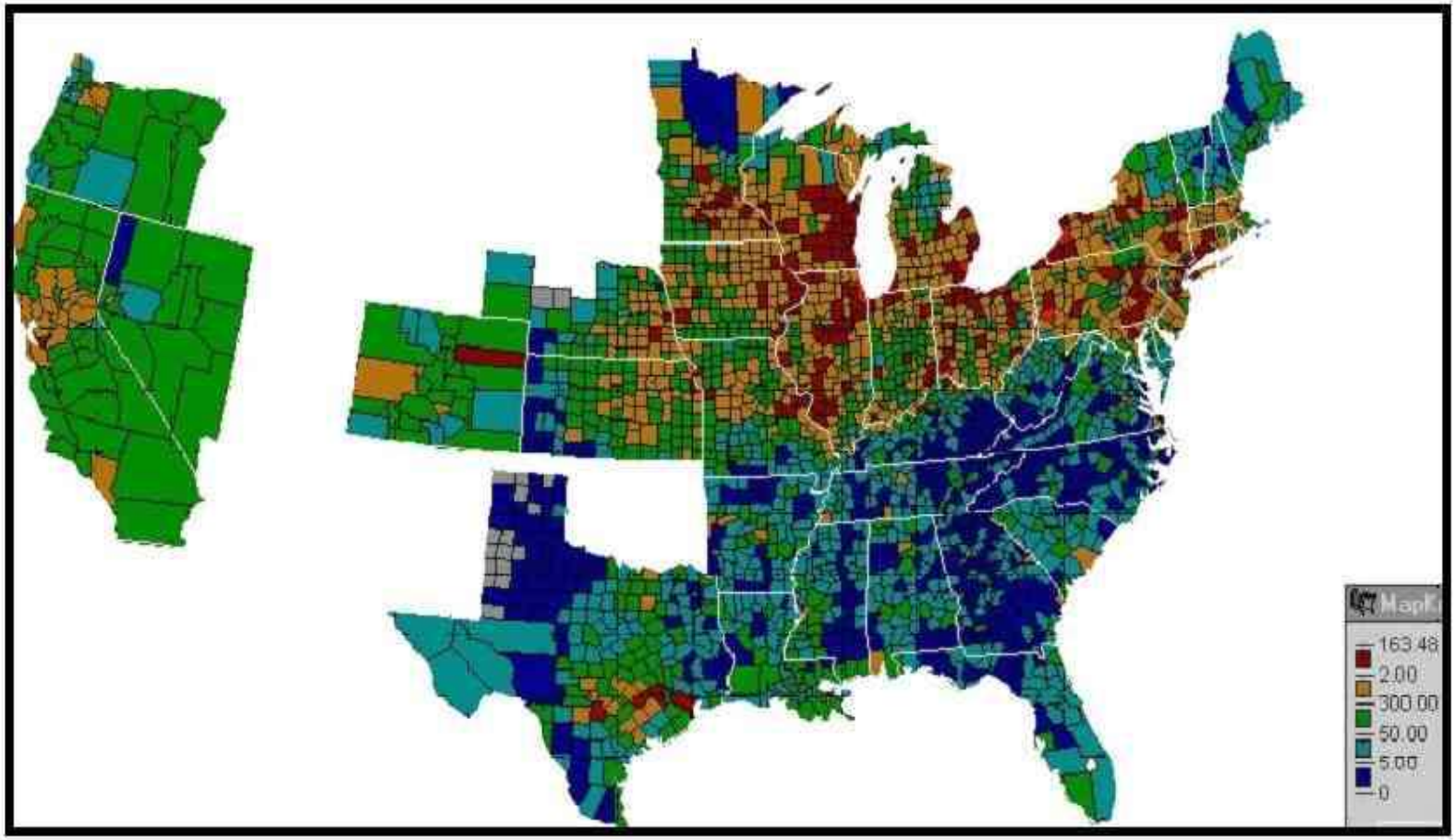
Non-Ashkenazi Jewish



Ashkenazi Jewish



Distribution of „Austrian“ immigrants from 1880s



Human Mutation 19: 575; 2002

Current Status of CF NBS (2007)

- Universally required
- Universally offered, but not required
- Offered to select populations or by request
- Required but not yet implemented
- Advanced planning stages
- Considering various options
- No information on current intentions

CDC

MMWR

Morbidity and Mortality Weekly Report

Recommendations and Reports — October 15, 2004 / Vol. 53 / No. RR-13

Newborn Screening for Cystic Fibrosis

Evaluation of Benefits and Risks and Recommendations for State Newborn Screening Programs

Image courtesy of Natus Medical Incorporated

INSIDE: Continuing Education Examination

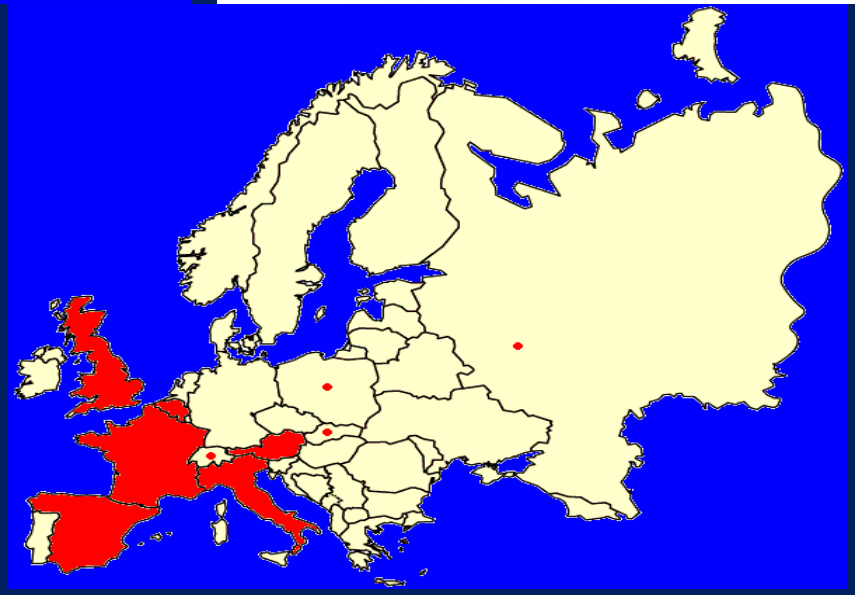
DEPARTMENT OF HEALTH AND HUMAN SERVICES
CENTERS FOR DISEASE CONTROL AND PREVENTION

Severe CF Malnutrition at Diagnosis

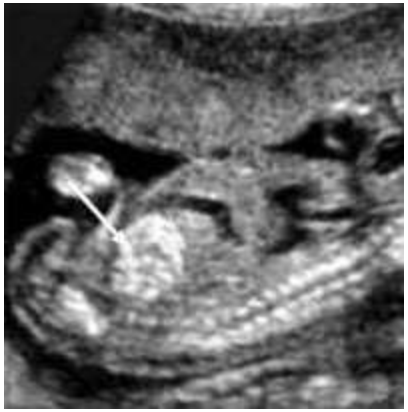
(3 month old diagnosed during 2001 in a nonscreening state)

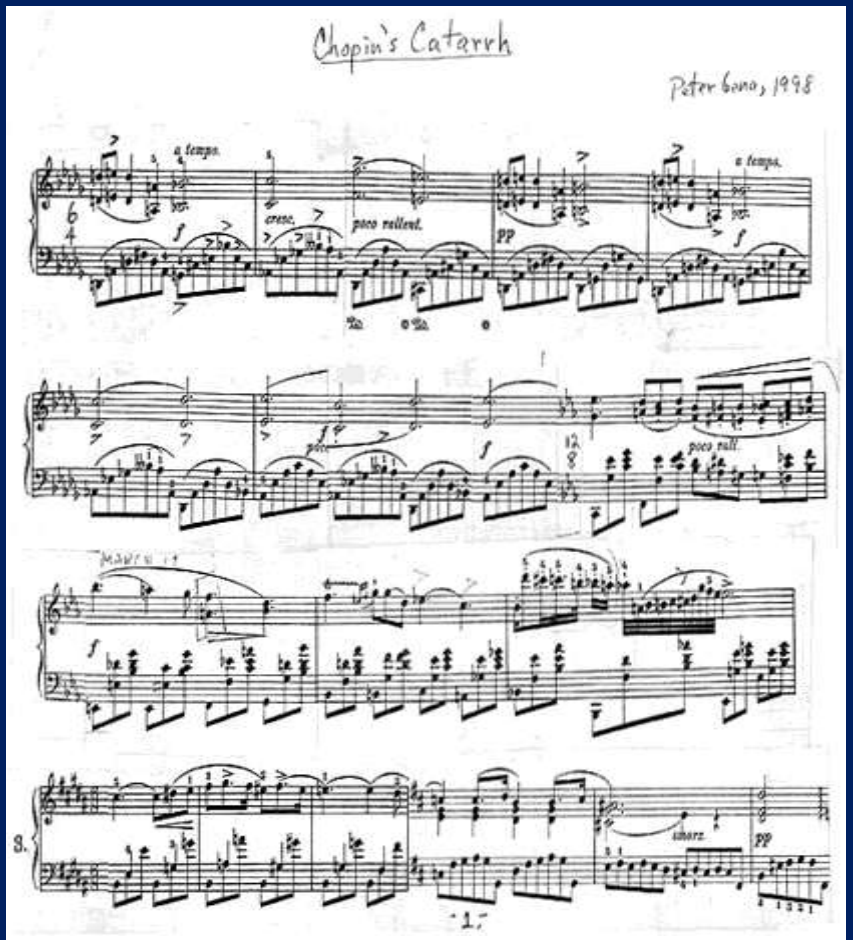
Potentially fatal protein-energy malnutrition with salt depletion

Photo courtesy of Frank J. Accurso, MD



Correlation between the incidence of CF and the frequency of the F508del mutation + influence of PND on CF incidence





Chopin's Catarrh (Nocturnes), is the first purely instrumental composition to come out of DNA research with geneticist, Charles Strom. Using an algorithm for converting DNA sequences into musical parameters, the cystic fibrosis sequence was transformed into musical notation.

DNA music

Musical Synthesis of DNA Sequences

Peter Gena, Ph.D., Charles Strom, M.D., Ph.D.

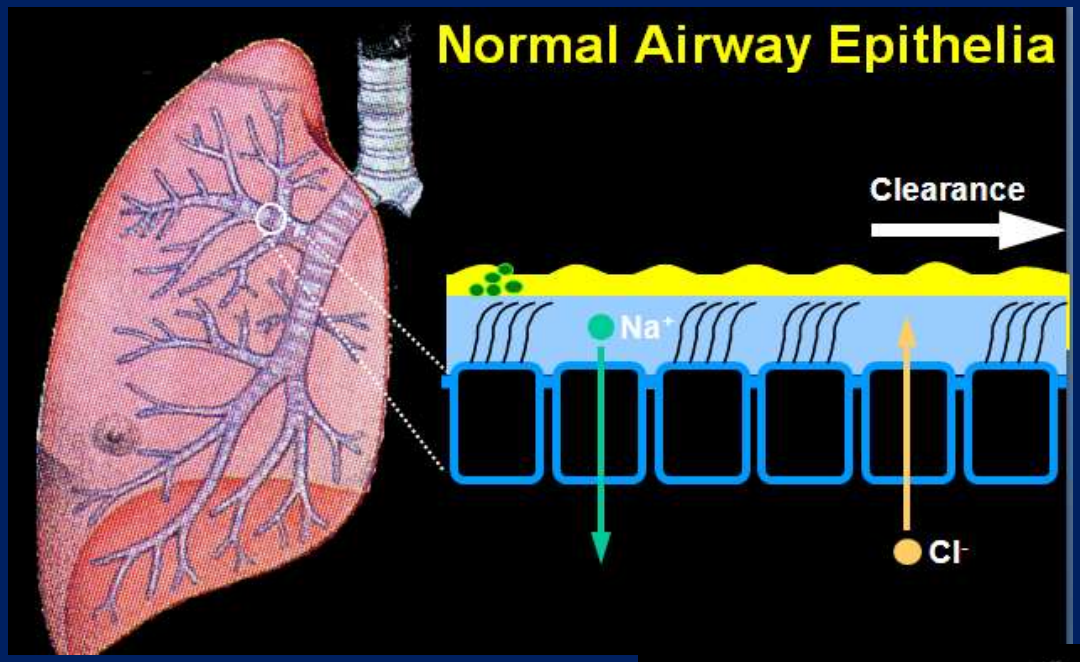
School of the Art Institute of Chicago, Illinois Masonic Medical Center
Chicago, Illinois USA

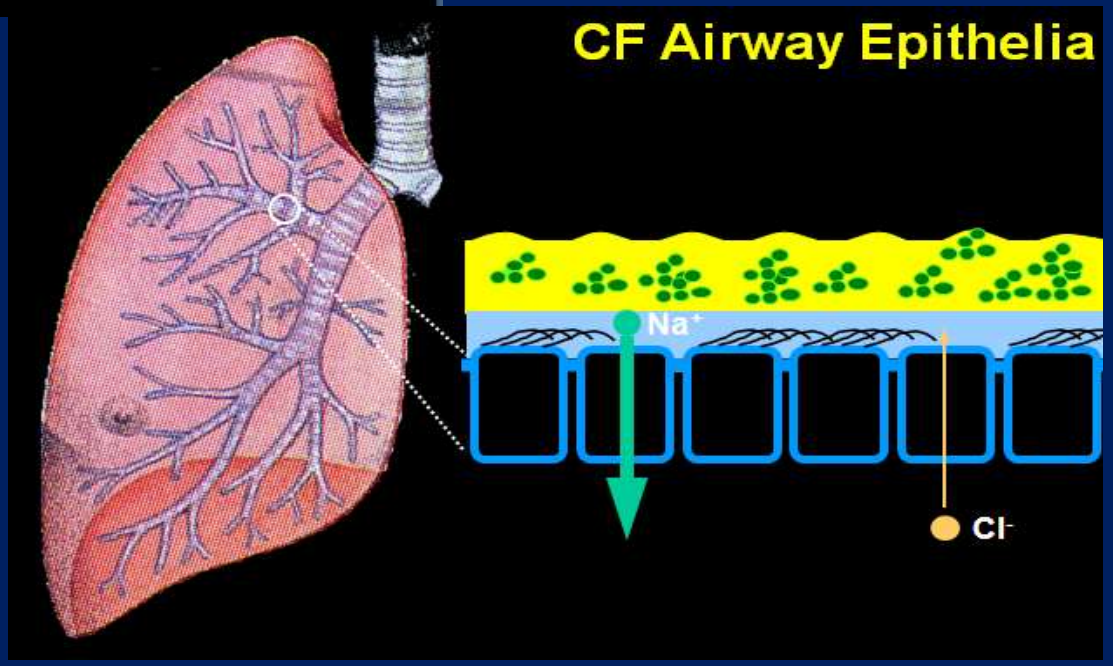
Abstract

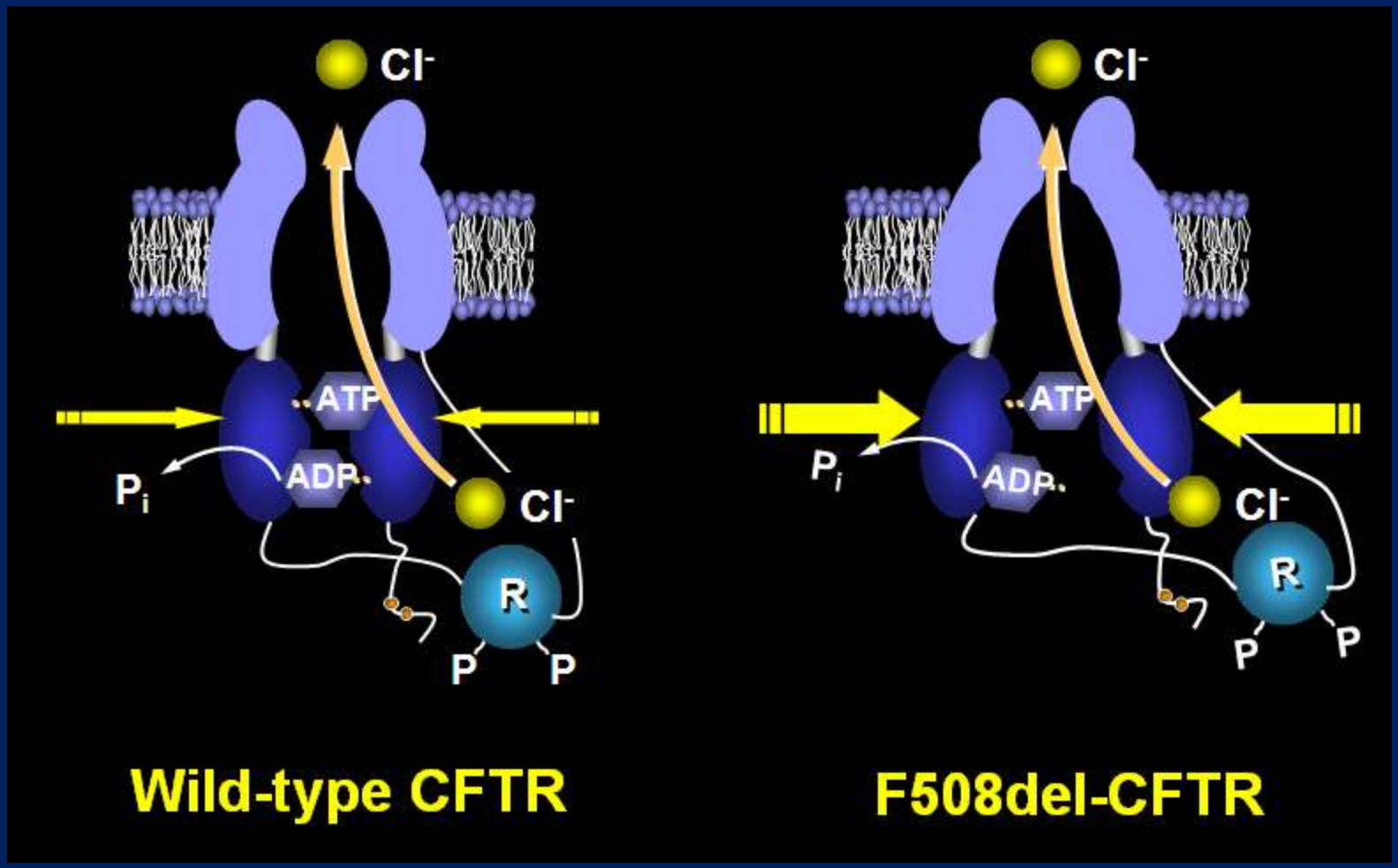
As a consequence of the Human Genome Project, there has been an explosion of primary DNA sequencing data available on CD ROM. This includes complete genomes of viruses, partial genomes of bacterias, and complete sequences for hundreds of human proteins. Consequently, we began to envision a type of computer-generated music that would take cues for its musical parameters directly from the physiological ones present in DNA. A DNA sequence consists of a specified order for the production of amino acids. The physical properties of amino acids (dissociation constant, molecular weight, and chemical class) combined with the properties of the individual bases (melting temperatures) provide the basis for inheritance and evolution and our musical compositions. The converted results, one for each codon, represent distinct musical actions in MIDI note events. Thus far, we have generated musical compositions from several human, viral, and bacterial sequences. This paper outlines our research.

XI Colloquio di Informatica Musicale, Univeristà di Bologna, 1995

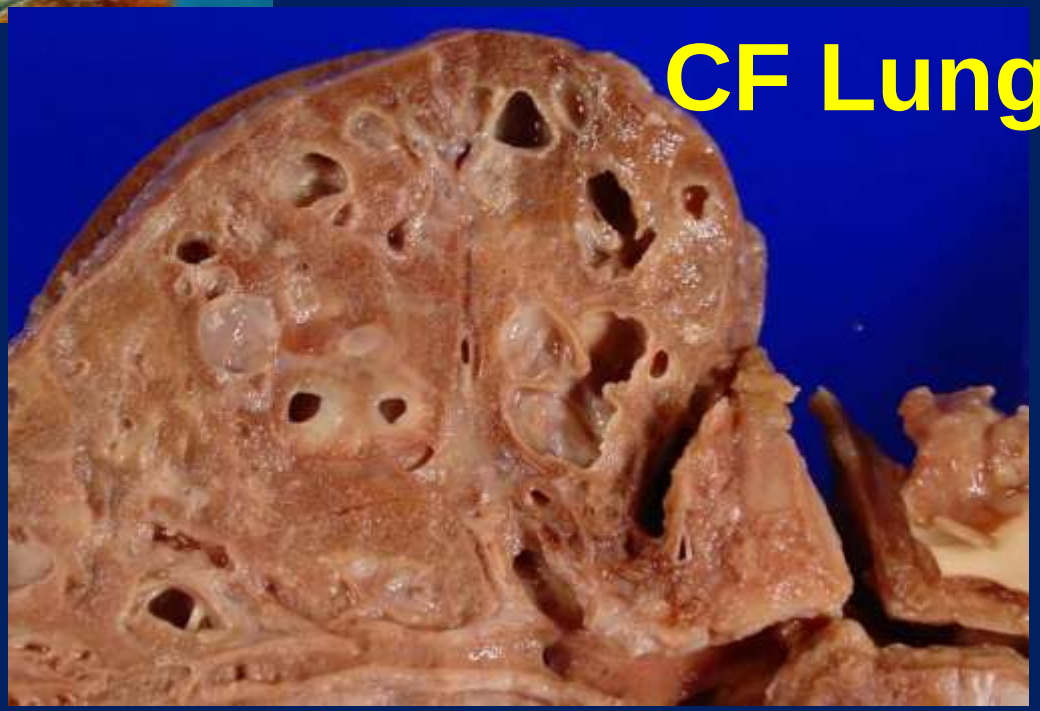
http://www.petergena.com/DNAmus.html


Normal Airway Epithelia
Clearance
Na+
Cl-


CF Airway Epithelia
Na+
Cl-

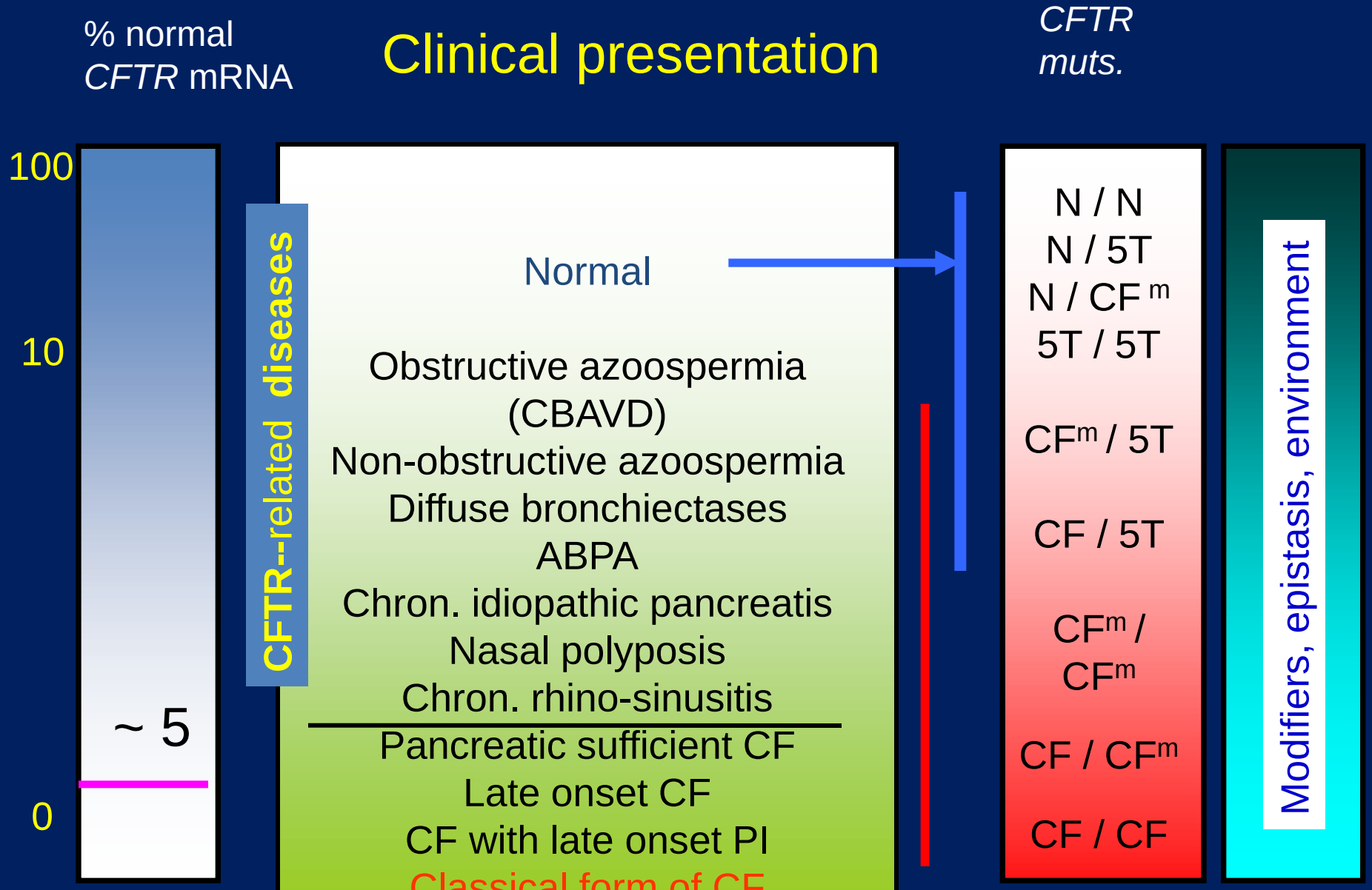

Cl-
ATP
ADP
Pi
Cl-
R
P
P
Wild-type CFTR
Cl-
ATP
ADP
Pi
Cl-
R
P
P
F508del-CFTR

Normal Lung

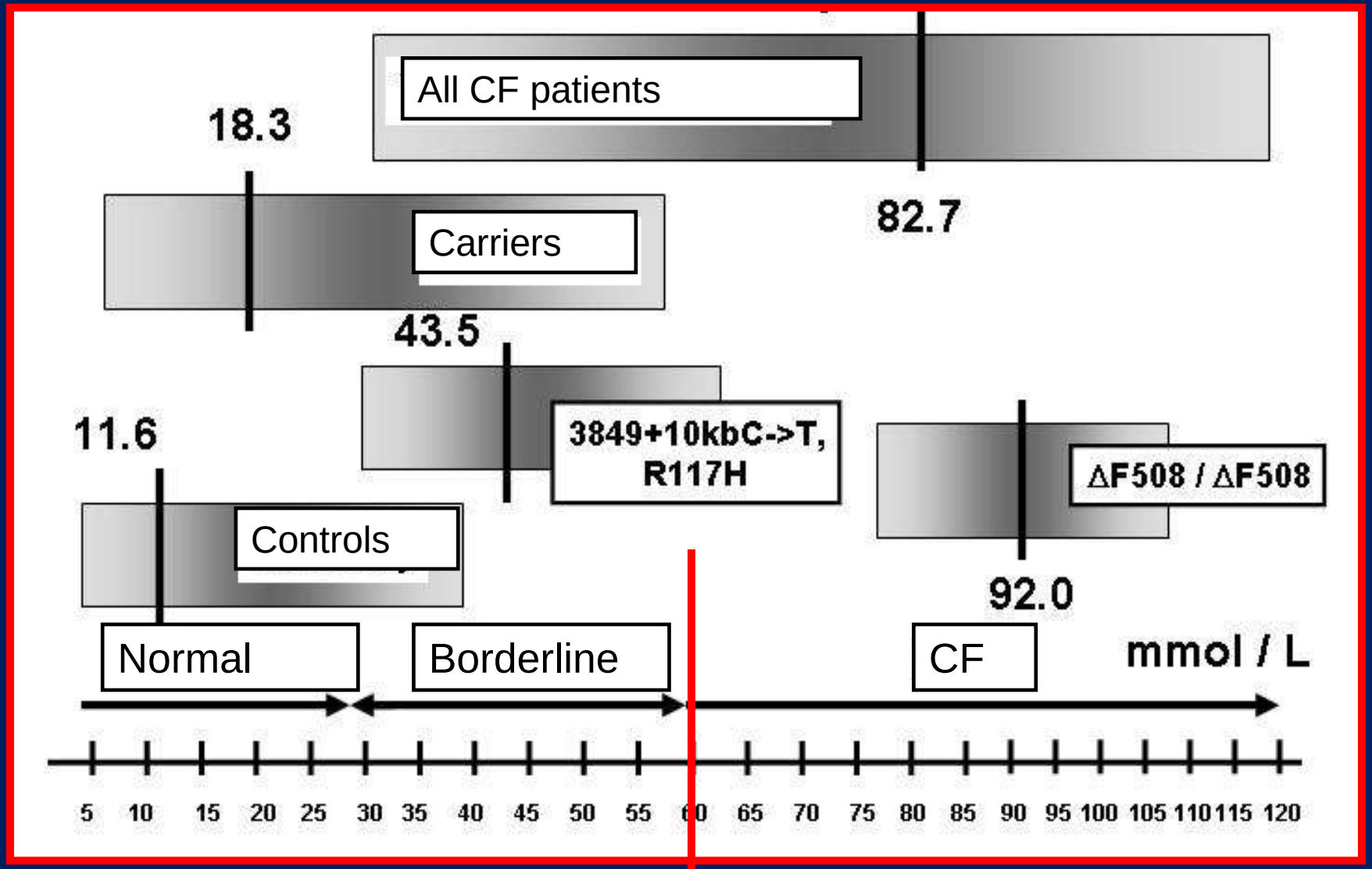


CF Lung

Phenotypes associated with *CFTR* alleles



Comparison of sweat Cl^- concentrations in Czech controls, carriers and patients



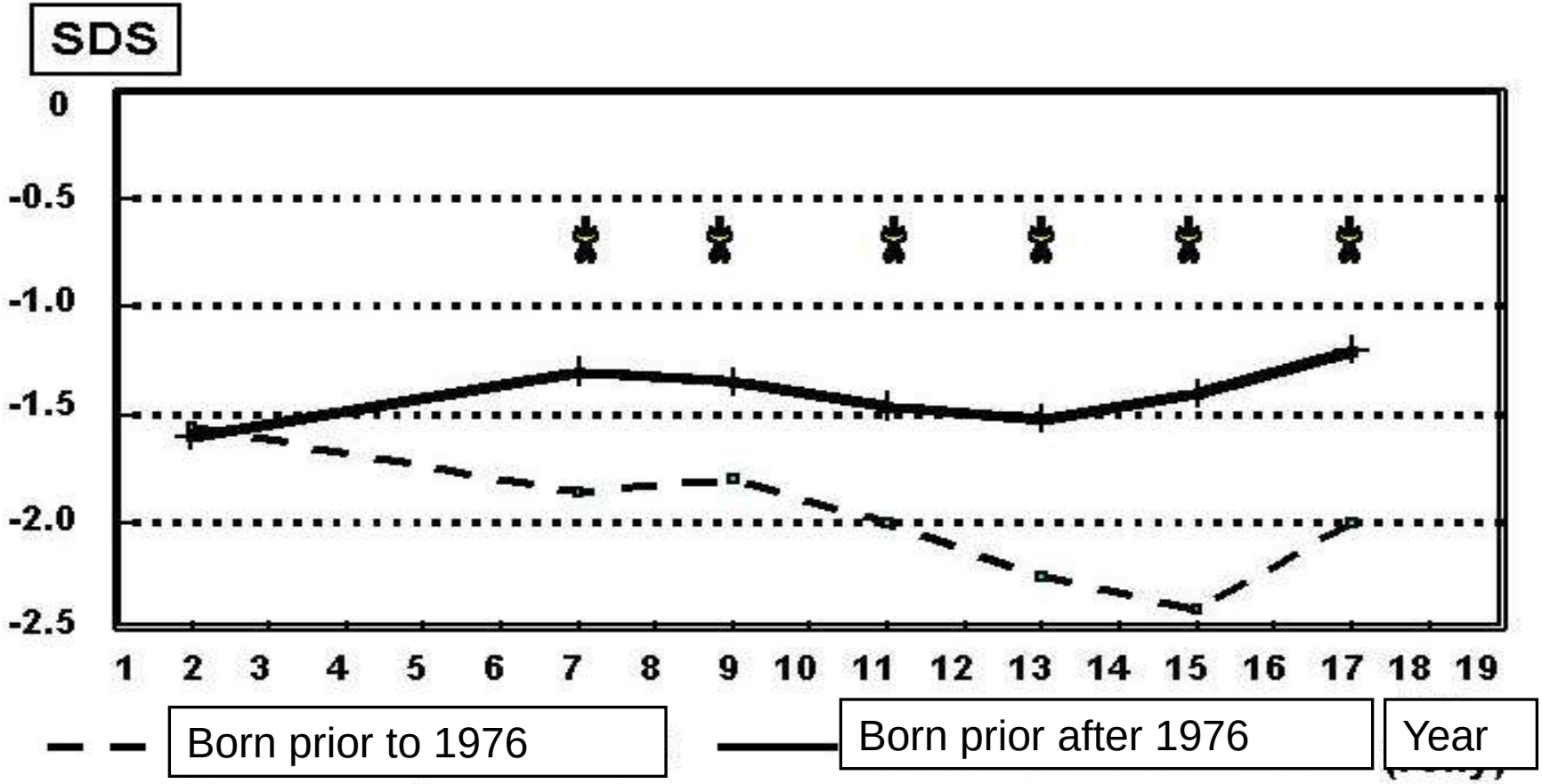
Pulmonary Manifestations

	African-American	Caucasian	p value
% Cultured	90.7%	91.3%	NS
P. aeruginosa	54.1%	59.8%	0.009
Klebsiella (sp.)	3.1%	1.3%	<0.001
Aspergillus (sp.)	3.5%	5.9%	0.02
FEV_1	71.7 ± 28.3	72.2 ± 28.7	NS
FVC	84.9 ± 25.1	85.1 ± 24.7	NS

Gastrointestinal Manifestations

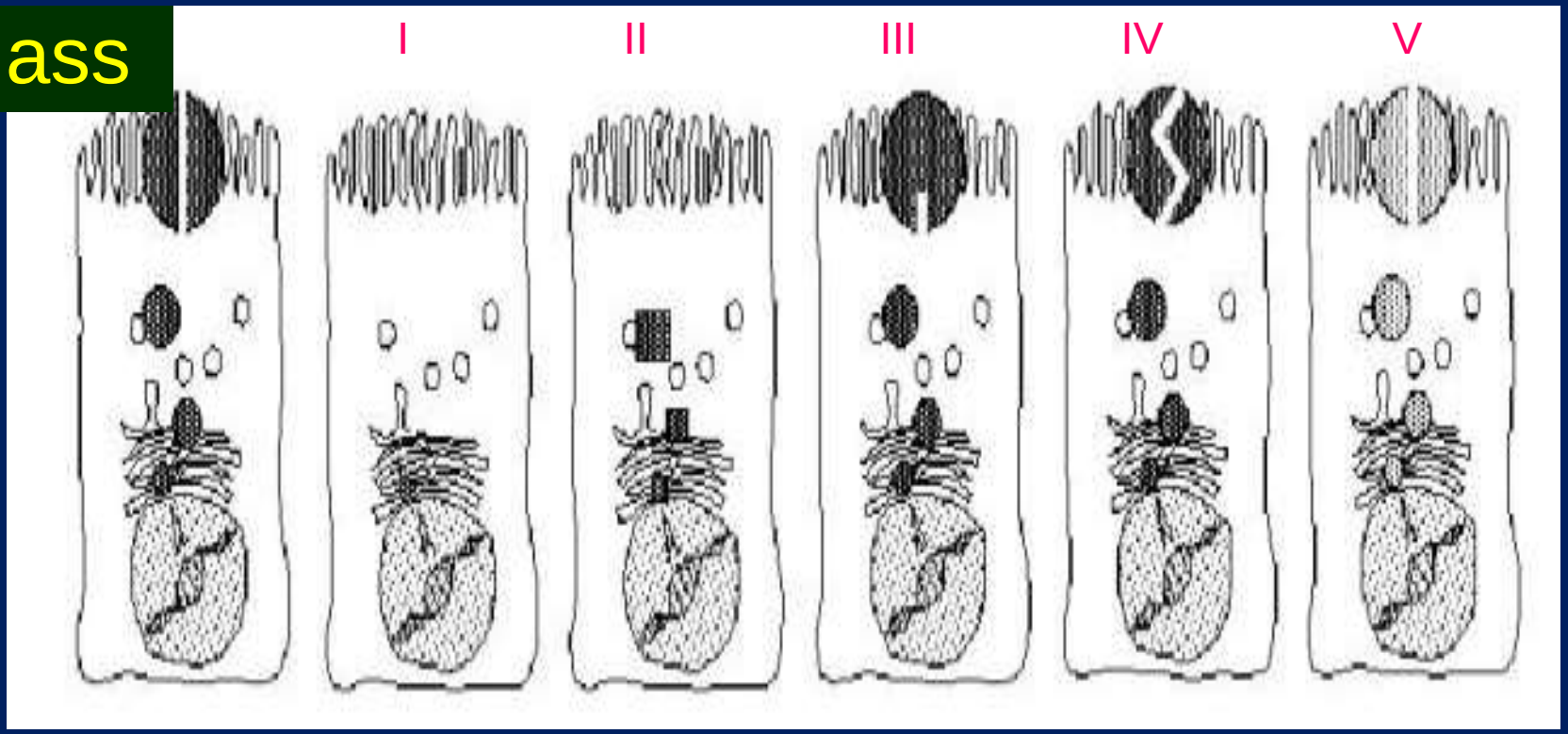
	African-American	Caucasian	p value
Pancreatic insuff.	95.2%	93.7%	NS
Meconium ileus ↓	12.7%	17.6%	< 0.001
DIOS ↑	4.5%	2.2%	< 0.001

Non-genetic modifiers: role of better therapy on mid-arm circumference (MAC) = nutritional s.



Vávrová et al. Čs-Pediat 54 : 220; 1999

Functional classification of *CFTR* alleles

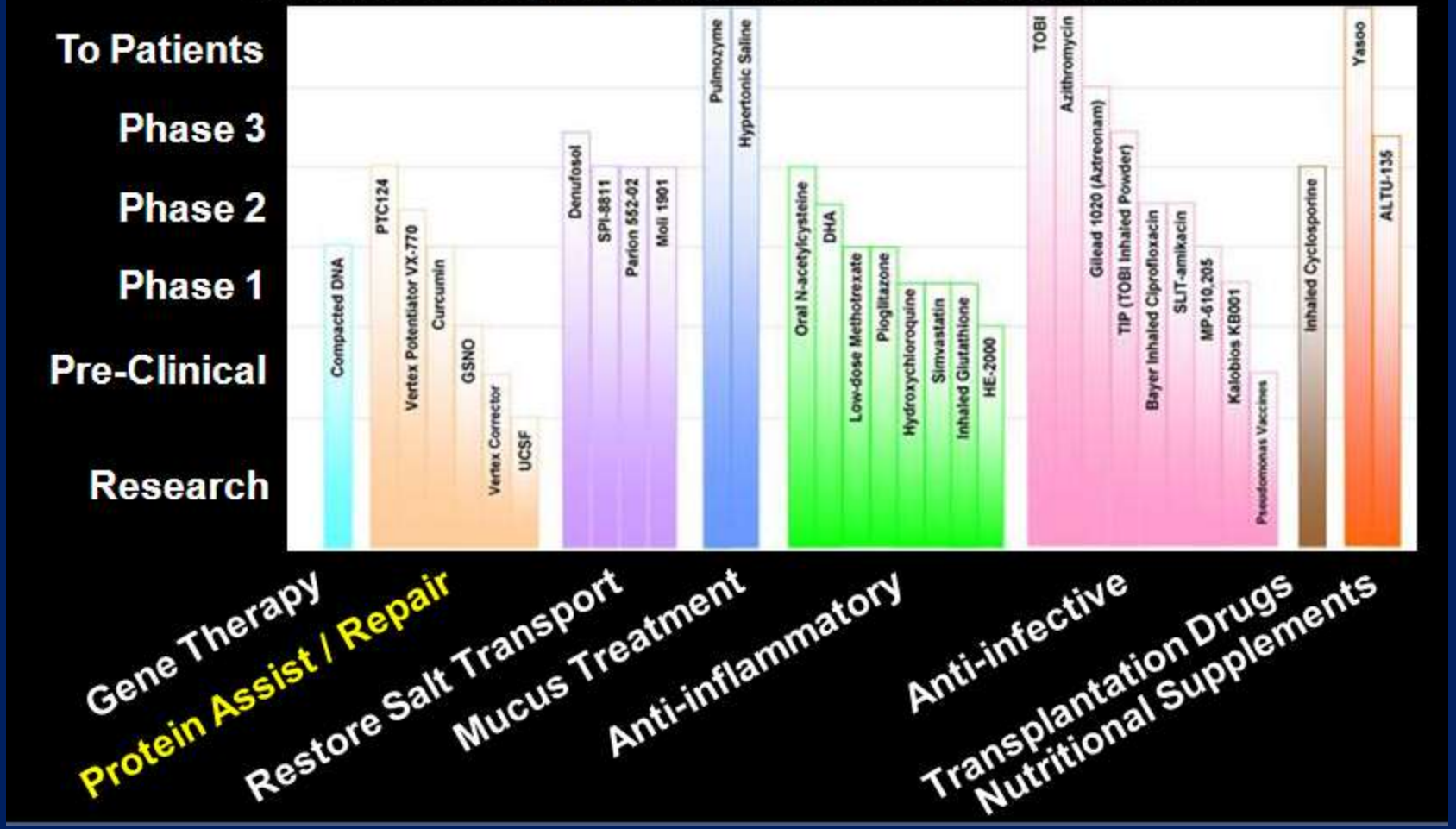


Representative example	G542X	F508del	G551D	R347P	3849+10kbC-T

Di = disturbance (qualitative) ; De = decrease (quantitative)

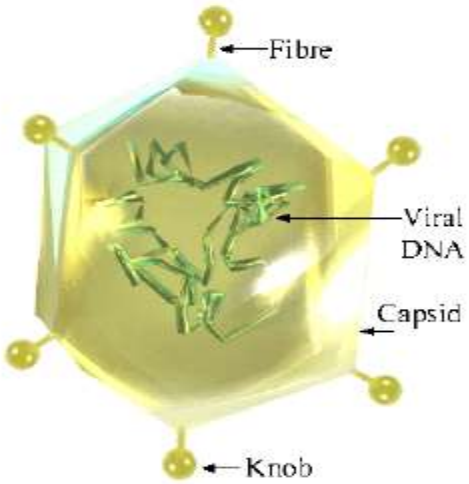
The CF Drug Development Pipeline

http://www.cff.org/research/DrugDevelopmentPipeline/



Gene therapy for CF

Adenovirus

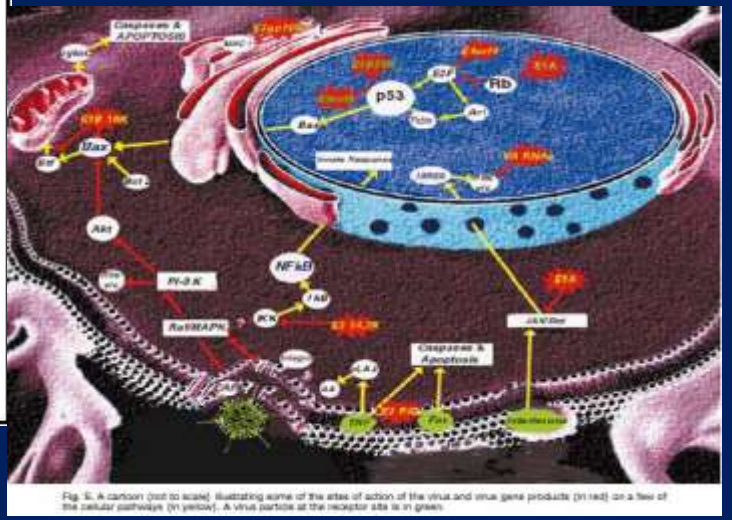


Advantages:

1. Efficiency of transduction is high
2. High level gene expression
3. Slightly increased capacity for exogenous DNA

Disadvantages:

1. Expression may be transient
2. Cell-specific targeting difficult to achieve
3. Virus uptake is ubiquitous
4. Safety



Adenovirus associated virus (AAV)

Advantages:

- not associated with disease
- can obtain high titered virus stocks (10^9- 10^{10}/ml)
- small genome that is easy to manipulate
- stable integration into the genome in chromosome 19
- infects both dividing and nondividing cells

Disadvantages:

- can only contain ~4.5 kb of DNA

In vitro - OK
Ex vivo - N/A
In vivo – problems

Gene therapy failure

Online NewsHour

a NewsHour with Jim Lehrer Transcript

ONLINE FOCUS

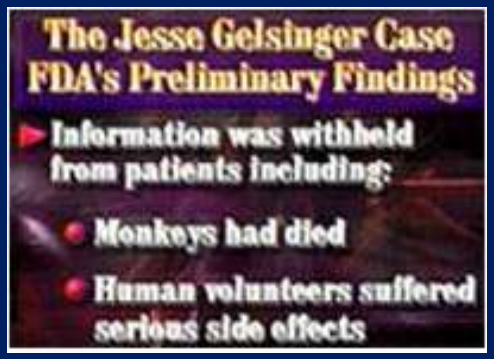
GENE THERAPY

February 2, 2000

As gene therapy trials come under fire for underreporting harmful side effects, Congress begins investigating the proper balance between regulation and research. Susan Dentzer reports on developments in controversial gene therapy treatments.

The Health Unit is a partnership with the Henry J. Kaiser Family Foundation.

Click here to listen to this segment in RealAudio



oversight mechanisms that are in place. There is absolutely no room, no place for mistakes that compromise patient safety.

American Society of Gene Therapy

Dr. W. French Anderson

A sudden breakthrough

SUSAN DENTZER: The death of the teenager, 18-year-old Jesse Gelsinger, who suffered from a rare liver disorder, occurred during an experiment at the university of Pennsylvania. Gelsinger was injected with special viruses designed to carry healthy copies of a gene into his body. His father, Paul Gelsinger, told Senators today that his son's disease was actually under control, but that he agreed to participate in the clinical trial to help other sufferers from the disease.

PAUL GELSINGER: Jesse was doing exceptionally well on his medications, and nothing should have prevented him from living a full and happy life. He believed, after discussions with representatives from Penn, that the worst that could happen in the trial would be that he would have flu-like symptoms for a week. He was excited to help.

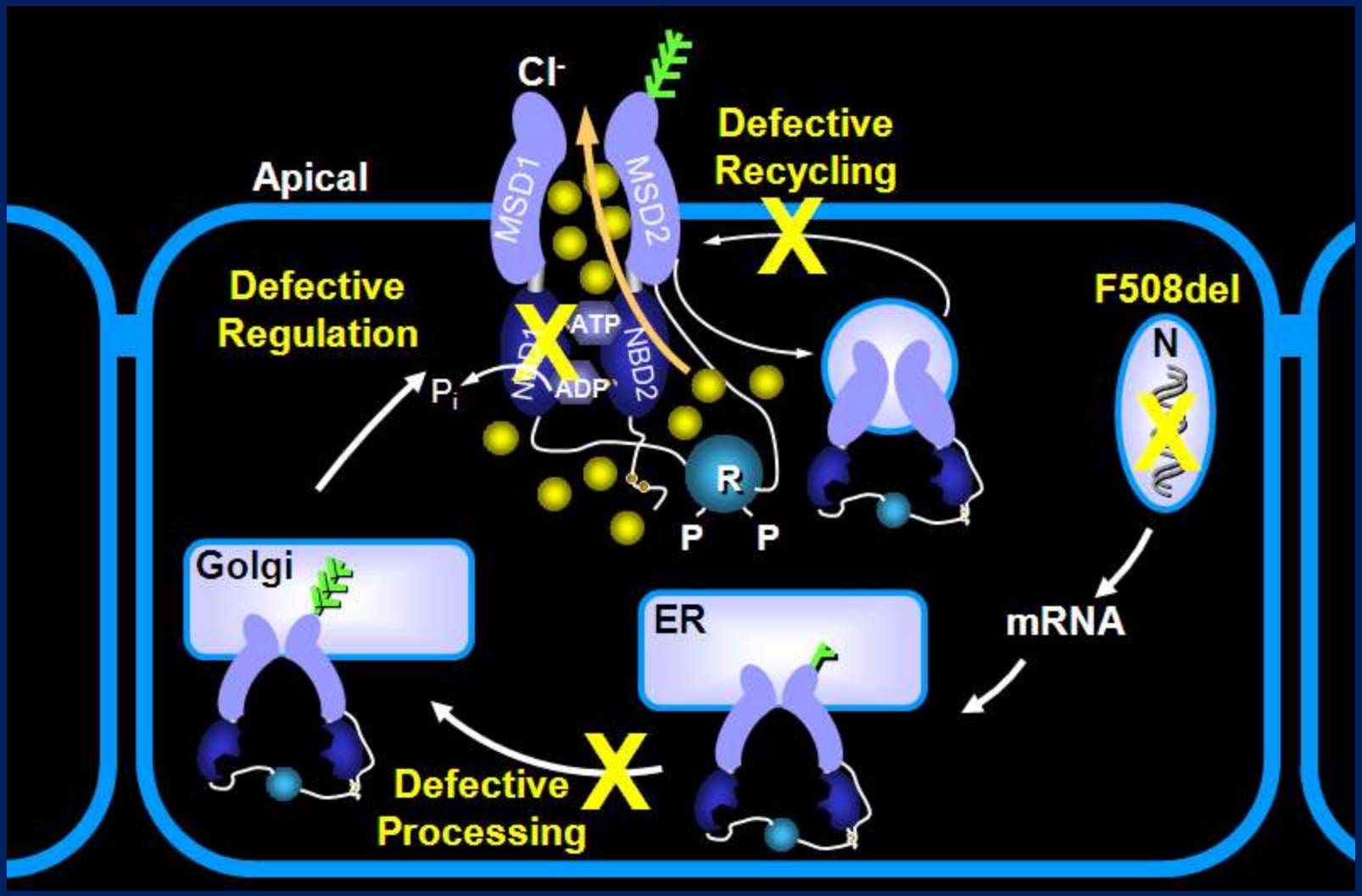
http://www.pbs.org/newshour/bb/health/jan-june00/gene_therapy_2-2.html

Drug Strategies to Rescue the Protein Folding Defect of F508del-CFTR

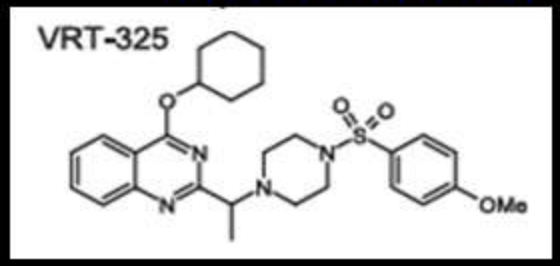
A variety of drug strategies are being evaluated to overcome the defective processing of F508del-CFTR. These drugs termed CFTR correctors include:

- Enhanced gene transcription (e.g. 4-phenylbuyrate)
- Calcium-pump inhibitors (e.g. thapsigargin)
- Chemical chaperones (e.g. glycerol, myo-inositol)

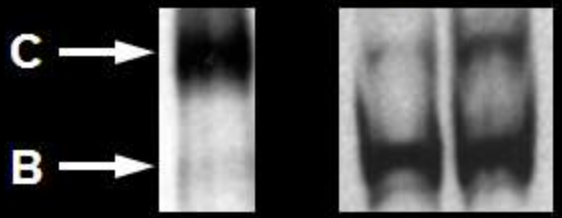
Other CFTR correctors (e.g. VRT-325) have been identified by high-throughput screening.


Cl-
Apical
MSD1
MSD2
Defective Recycling
Defective Regulation
ATP
ADP
NBD2
Pi
R
P
P
F508del
N
Golgi
ER
mRNA
Defective Processing

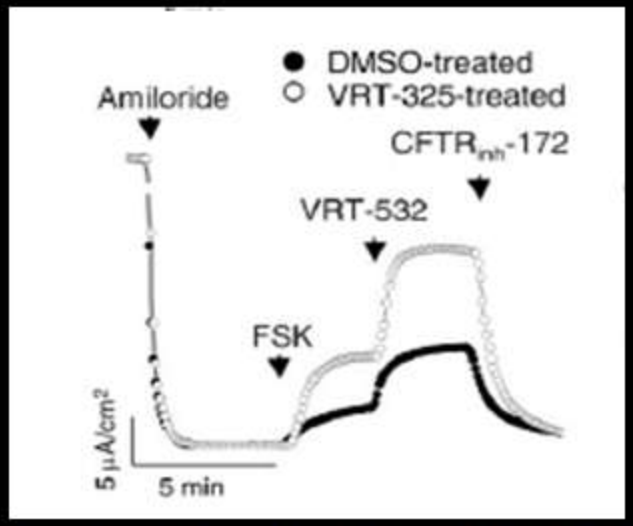
VRT-325 Rescues the Cell Surface Expression of F508del-CFTR in Human Bronchial Epithelia



wt-HBE F508del-HBE



DMSO
VRT-325



Drug treatment: 6.7 μM for 48 h

From: Van Goor, F. *et al. Am. J. Physiol.* 290, L1117-L1130, 2006

Gentamicin-Induced Correction of CFTR Function in Patients with Cystic Fibrosis and CFTR Stop Mutations

Michael Wilschanski, M.D., Yaacov Yahav, M.D., Yasmin Yaacov, B.Sc., Hannah Blau, M.D., Lea Bentur, M.D., Joseph Rivlin, M.D., Micha Aviram, M.D., Tali Bdolah-Abram, M.Sc., Zsuzsa Bebok, M.D., Liat Shushi, M.Sc., Batsheva Kerem, Ph.D., and Eitan Kerem, M.D.

2003

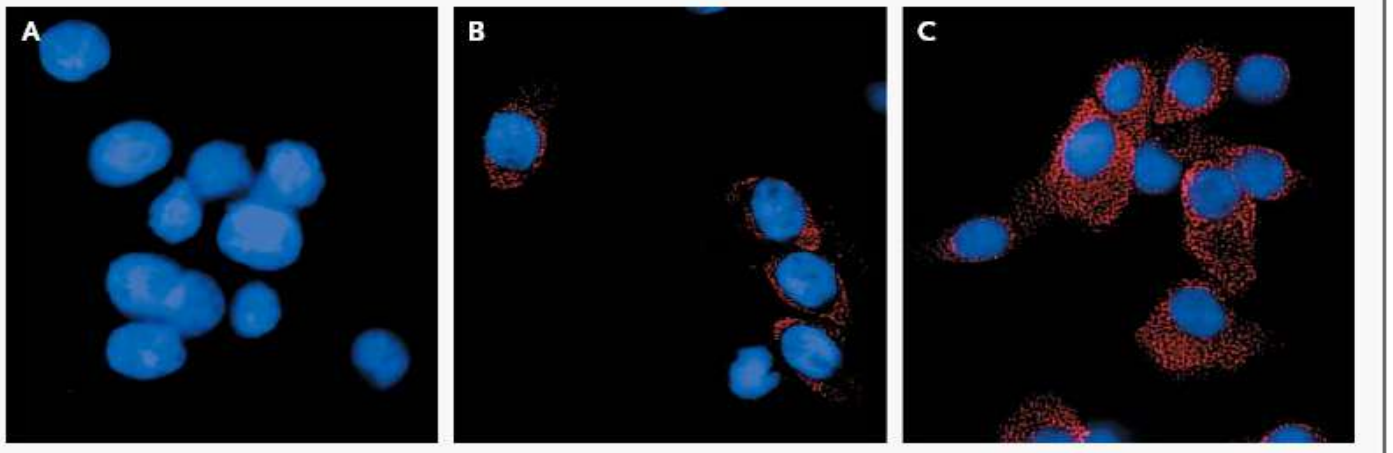


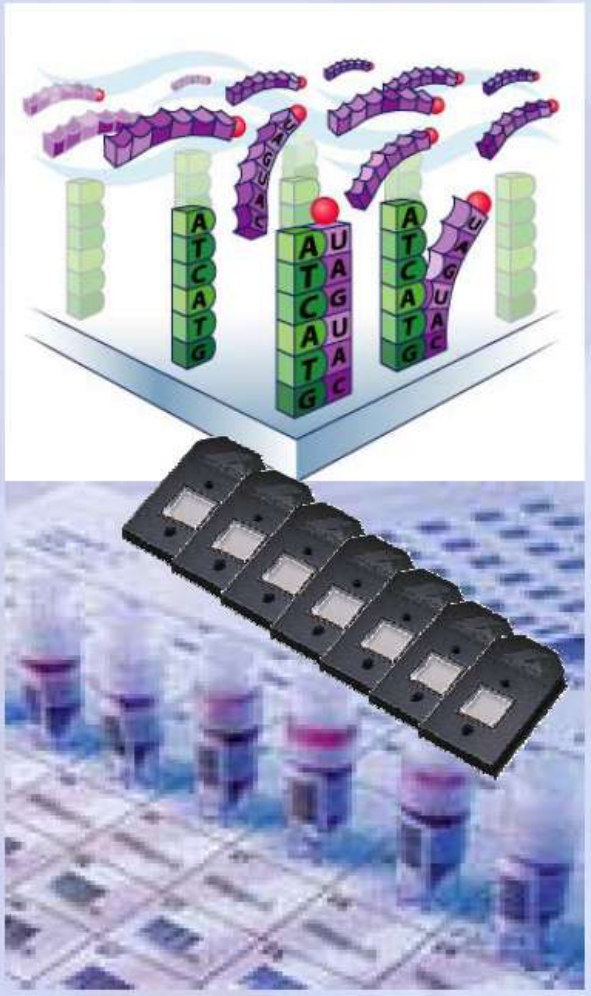
Figure 4. An Example of Immunostaining for the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) in a Negative Control Involving Nonimmune Mouse CFTR IgG (Panel A) and in a Patient with the W1282X/ΔF508 Mutation before (Panel B) and after (Panel C) Gentamicin Treatment (×100).

The CFTR-positive cells have a peripheral and surface pattern of staining after gentamicin treatment.

In patients with cystic fibrosis who have premature stop codons, gentamicin can cause translational "read through," resulting in the expression of full-length CFTR at the apical cell membrane, and thus can correct the typical electrophysiological abnormalities caused by CFTR dysfunction

Genomics based Medicine

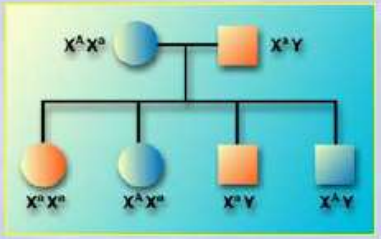
- Medicine: From Descriptive to Experimental Data driven Science -



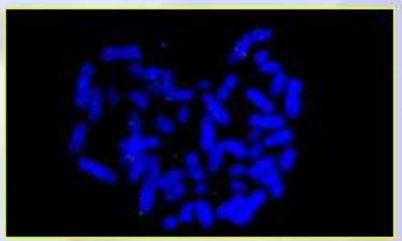
Forensics

Clinical Trials, SNPs

Epidemiological Studies



Genotyping



Cytogenetics

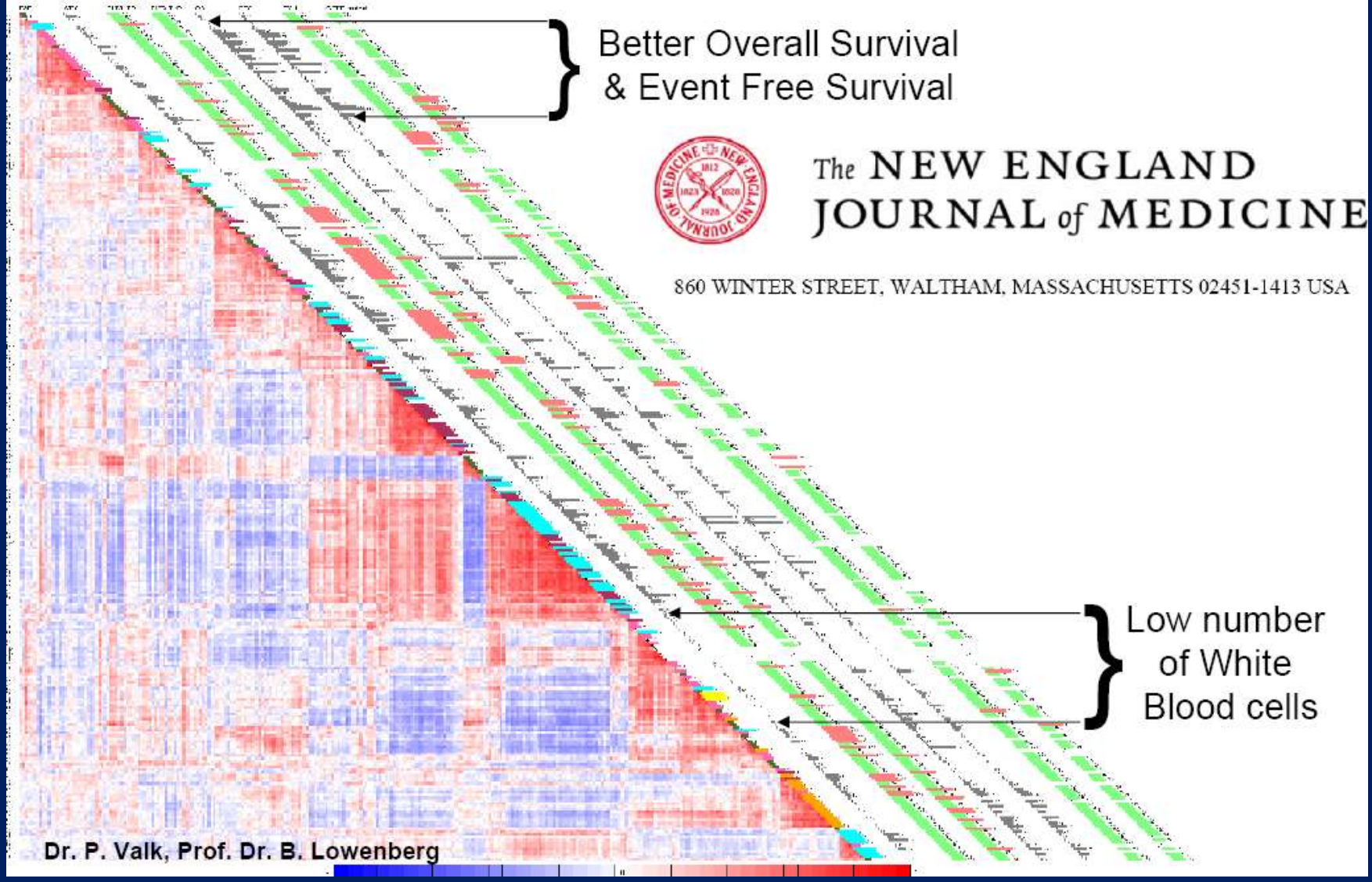
Individualized Medicine

Basic Research

Human Diagnostics

Leukemia Patient Classification

- Genotype-Phenotype Correlation -



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860 WINTER STREET, WALTHAM, MASSACHUSETTS 02451-1413 USA

Dr. P. Valk, Prof. Dr. B. Lowenberg

“Lifescience Data Explosion”

„Mind the GAP“

23andMe genetics just got personal.

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1866: Gregor Mendel discovers the laws of inheritanc

200,000 years ago: *Homo sapiens* walks the Earth. 2003: The Human Genom

2007: 23andMe introduces the first Personal Gen

Unlock the secrets of your own DNA. To

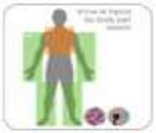
Welcome to 23andMe, a web-based service that helps you read and understand your DNA. After providing a saliva sample using an at-home kit, you can use our interactive tools to shed new light on your distant ancestors, your close family and most of all, yourself.

Sign up for our Email Newsletter

news **What's new at 23andMe**

Feb 21, 2008: Personal Genome Service enhanced with more Gene Journal content, a new Paternal Ancestry feature, and free demo accounts.

Gene Journal

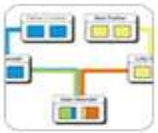


What do your genes say about you?

Ancestry

Who were your ancient ancestors?

Family Inheritance



Do you have your mother's sense of taste?

Genome Labs

Would you like to search your genome?

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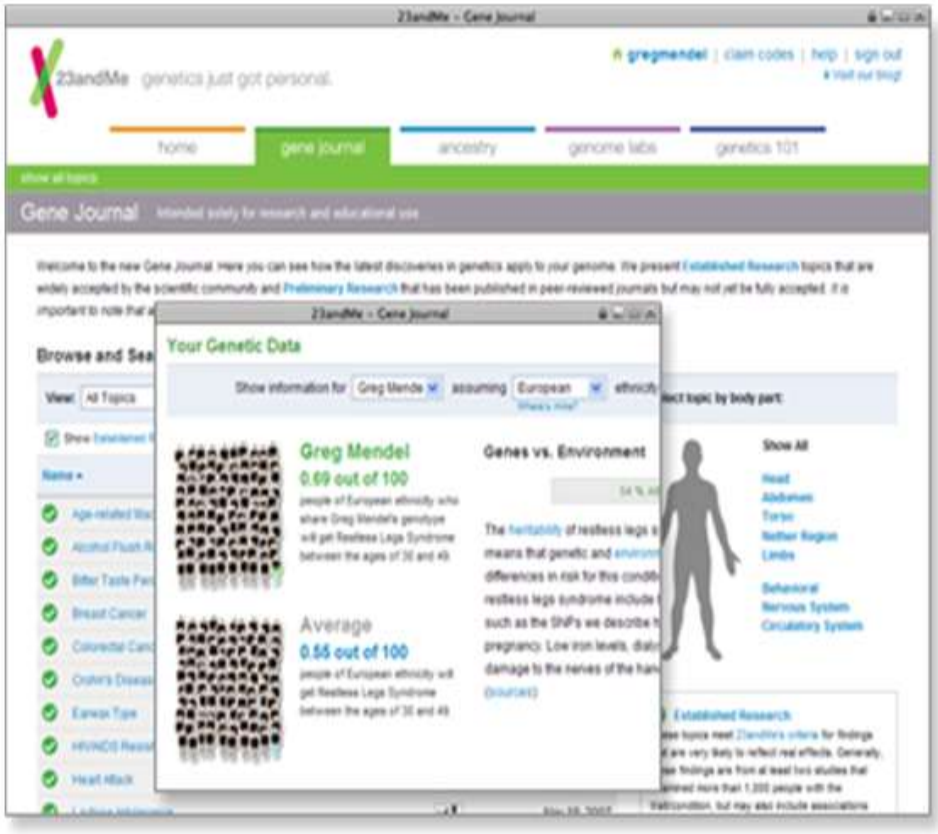
Prohledat Web | Stránky pouze česky

Reklamní programy - Všechno o Google - Google.com in English

Chat: Linda Avey and Anne Wojcicki

www.23andme.com

Take a look at the odds.



The 23andMe Odds Calculator

The 23andMe Odds Calculator helps you put it all in perspective, allowing you to combine genetic information, age, and ethnicity to get an idea of which common health concerns are most likely to affect a person with your genetic profile. While the Odds Calculator is neither a medical diagnostic nor a substitute for medical advice, it can help you confront the bewildering array of health news reported in the mass media and help you decide where you may want to focus your attention.

At Illumina, a San Diego biotech firm, chips are prepared for genotyping in the "decoding bay." [1]

Mission Statement

23andMe's mission is to be the world's trusted source of personal genetic information.

All of us have 23 pairs of chromosomes

17% of us are left-handed

88% of us have wet earwax

None of us are world-class sprinters

Navigenics™

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Your genes offer a road map to optimal health

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We're in the midst of the genomics revolution, discovering new connections between your DNA, your environment and your personal health and wellness. You can feel confident choosing Navigenics to help you harness this information, because:

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- Our lab is certified under CLIA, the law covering accuracy and timeliness of test results.
- Our genetic counselors, available by phone, help you interpret your results.
- Our medical partnerships help inform and guide your next steps on the way to optimal health.
- Our practices are consistent with HIPAA standards for privacy and protection of your personal data.

In the news

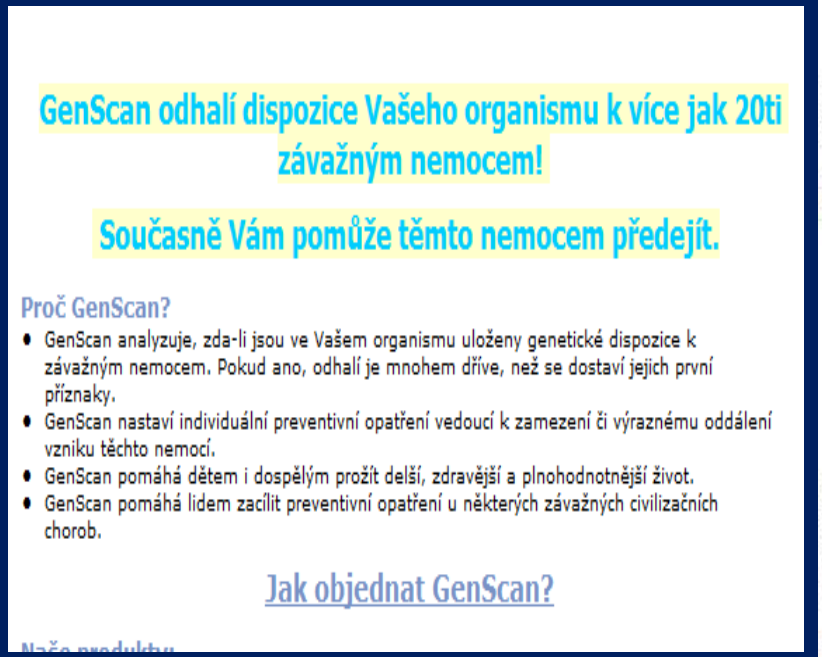
Fast Company: Are There Holes In My Genes?
A leading industry reporter explores the genetic testing market and captures her personal experience with Navigenics Health Compass. Read more

Navigenics Launches with Preeminent Team of Advisers,

www.navigenics.com

Filosoficko- sociální problém:

„Právo vědět“
¨“Svoboda podnikání“
„Svoboda volby“
„Socialistické regulace“



sgpph

Society for Genomics Policy and Population Health

Genetic Susceptibility Testing

Spring Event: Thursday 8 May 2008 10.00–16.30
Education Centre, Birmingham Women's Hospital

The last year has seen tremendous progress in our understanding of the genetics of common diseases. Recent findings from the new wave of genome-wide association studies are being rapidly translated into commercial testing services in Europe and the United States, with many new tests being offered direct-to-consumer over the internet. This one-day conference will review scientific progress in this area and consider the policy issues raised by this new generation of susceptibility tests, with a particular emphasis on the regulation of consumer genetics.

The Society for Genomics Policy and Population Health (SGPPH) aims to promote interdisciplinary discussion and research into issues raised by developments in genomics and health.

LG SP
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GHC

Certifikované semináře pro lékaře pořádané GHC Genetics ve spolupráci se Společností pro lékařskou a prediktivní genetiku

Nové možnosti prediktivní genetiky v lékařské praxi
Termín školení pro lékaře bude upřesněn v následujících dnech.
Máte-li zájem se stát našim certifikovaným lékařem, můžete se níže přihlásit do certifikačního programu.

The NEW ENGLAND JOURNAL of MEDICINE

Perspective

JANUARY 10, 2008

Letting the Genome out of the Bottle — Will We Get Our Wish?

David J. Hunter, M.B., B.S., Sc.D., M.P.H., Muin J. Khoury, M.D., Ph.D., and Jeffrey M. Drazen, M.D.

It may happen soon. A patient, perhaps one you have known for years, who is overweight and does not exercise regularly, shows up in your office with an analysis of his whole genome at r

single-nucleotide polymorphisms (SNPs). His children, who were concerned about his health, spent $1,000 to give him the analysis as a holiday gift. The test report states that his genomic profile is consistent with an increased risk

types. These studie
croarrays that can a
or more SNPs in ea
ple; researchers u
croarrays to examin
al differences in inh
variability and to

The test undergone by the patient described above is one of the products of this new knowledge.

As of November 2007, two

NEJM 2007-2008

PERSPECTIVE

The Mixed Promise of Genetic Medicine

Carl Elliott, M.D., Ph.D.

In the early decades of the 20th century, most Americans considered cosmetic surgery to be just a few steps removed from quackery. Many observers saw the desire for cosmetic surgery as a mark of vanity, and physicians tended to believe that such surgery violated their ethical injunction to do no harm. Yet by the end of the century, cosmetic surgery had become a multibillion-dollar business, and it is now an accepted part of mainstream medicine, with its own professional journals and associations. Cosmetic-surgery clinics are sponsored by elite academic centers such as Stanford, Johns Hopkins, and the Mayo Clinic. Even by medicine's move into the consumer marketplace. Physicians today prescribe drugs to lengthen attention spans, strengthen erections, and smooth out wrinkled brows, even when they are not entirely convinced that what they are treating is a medical need rather than a consumer desire. Many others write prescriptions for conditions that blur the boundary between pathology and ordinary human variability: synthetic growth hormone for idiopathic short stature, antidepressants for social anxiety disorder, and hormone-replacement therapy for the effects of menopause. The line between what consumers want and have a deaf child by seeking out a sperm donor with five generations of deafness in his family. Could such cases represent a glimpse into our genetic future?

Many of us feel uneasy about such a future, without being quite able to say why. Michael Sandel's graceful and intelligent new book, *The Case against Perfection*, is an extended effort to diagnose that unease. What troubles Sandel is not so much the particular kinds of traits and abilities that consumers might choose (deafness rather than super-hearing, for example) or even the possibility that these procedures will be bought and sold in the marketplace. It is the

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More Recently: Soda pop 20% of kids' calorie intake

DNA-based diet plan dubbed waste of money

NBC San Diego | Read article

WBNS TV | Read article

Nutritionist Carolyn Katzin is marketing a so-called "nutrigenomics" diet kit, in which you fill out a family history questionnaire and return some swabbed DNA samples for analysis. In return you receive a printed diet and nutrition plan.

However, a weight-loss specialist, a genetic researcher, and a spokesman for the National Cancer Institute all questioned the utility of such a method. Even Katzin doesn't seem to want to support the DNA claims too strongly, justifying the diet kit on "motivational" grounds.

Three reasons not to buy the NicoTest™ genetic test

GeneWatch UK

December 2004

What is the NicoTest™?

NicoTest is a new genetic test kit being marketed directly to the public via the internet (www.nicotest.com). It is marketed by a company called g-Nostics Ltd which hopes to sell it more widely in the future, both "over the counter" and via doctors in the National Health Service. G-Nostics is a "spin out" company from Oxford University. The university is a shareholder in the company and the test is based on research by Dr Robert Walton in the university's Department of Clinical Pharmacology[1]. Dr Walton is Chief Scientific Officer, Lead Inventor and co-founder of the company[2].

NicoTest is a smoking cessation programme, which includes three things:

1. A questionnaire and DNA test which the company claims will identify which smokers are more likely to respond to nicotine replacement therapy (nicotine patches, gum and inhalers) and less likely to respond to drugs such as bupropion (Zyban) when they try to quit smoking;
2. A second DNA test which the company claims gives the customer's metabolic profile (how their body responds to nicotine) and therefore how much nicotine replacement to take;
3. A programme of support to quit, including email, chat rooms and a computer programme (based on cognitive behavioural therapy).

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News › Science

Gene tests from shops and internet 'waste of money'

- No evidence that they work, say scientists
- Family history 'better indicator' of disease risk

Ian Sample, science correspondent
The Guardian, Monday January 30 2006
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UK news

A range of genetic tests sold in health shops and over the internet have been been branded a waste of money by leading scientists, because there is no evidence they work.

Gene tests claiming to measure a person's risk of developing intractable diseases such as cancer and Alzheimer's are being rushed to market even though studies to prove they benefit patients have not been done, researchers told a meeting on genomics and public health held by the Royal College of Physicians in London.

The tests differ from those available on the NHS, which generally look for single gene mutations known to cause disease, such as cystic fibrosis. The new tests are being developed to look for sets of genes scientists have linked, often tenuously, to a person's susceptibility to disease.

"Even if there's theoretical evidence the genes are linked to a disease, that's often far too little to go on. There's not one shred of evidence that these tests benefit human health," said Ron Zimmern, director of the public health genetics unit at Cambridge University.

Muin Khoury, director of genomics and disease prevention at the prestigious Centres for Disease Control and Prevention in Atlanta, told the meeting: "There are more than 1,000 genetic tests on the market now, with many available over the internet, but suffice it to say we have no idea whether they are of any value." In many

http://www.guardian.co.uk/science/2006/jan/30/genetics.research

Goetz P.
Předseda Společnosti lékařské genetiky ČLS JEP, Ústav biologie a lékařské genetiky 2. LF UK
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Místopředsedkyně Společnosti lékařské genetiky, Masarykův onkologický ústav Brno
Macek M.
Člen výboru Společnosti lékařské genetiky, Ústav biologie a lékařské genetiky 2. LF UK
Brdička R.
Člen výboru Společnosti lékařské genetiky, Ústav hematologie a krevní transfuze Praha
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Franková V.
Ústav pro humanitární studia v lékařství, 1. LF UK, Praha
Hořínová V.
Oddělení lékařské genetiky, Jihlava
Payne J.
Předseda Společnosti lékařské etiky, Ústav pro humanitární studiea v lékařství 1. LF UK
Hach P.
Předseda etické komise Mzd ČR

Hrozí zneužití – diskreditace lékařské genetiky

Souhrn: Lékařská genetika má bezesporu pohnutou historii, je poznamenána obdobími, kdy byla tragicky zneužita. Poznatky o genetické podmíněnosti normálních i abnormálních znaků, spolu s eugenickým hnutím byly v 30. a 40. letech min. století zneužity v nacistickém Německu v podobě rasové teorie a následné otřesné praxe rasové hygieny. Komunistické Rusko německé rasistické a eugenické aktivity odmítlo, ale zároveň odmítlo a likvidovalo v období lysenkoismu (50. léta) genetiku jako takovou.
Tato mementa je třeba stále mít na paměti, zejména nyní, kdy neustále se rozšiřující znalosti o lidském genomu přinášejí nové možnosti jak pozitivního medicínského využití, tak i zneužití. Existují vážné obavy z použití vědecky a klinicky nepodložených postupů. Navíc může dojít k závažnému zanedbání základních etických principů medicíny a k porušení Hippokratovy přísahy.

The Patient Perspective: Treatment Burden

I. Stenzel
Photo by Derek Powazek

http://www.thebreathingroom.org/

!!! Děkuji za pozornost !!!