The Novo Nordisk Foundation
Center for Protein Research

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Clinical Phenotyping and recording of medical data

Extraction of disease phenotypes from electronic patient records

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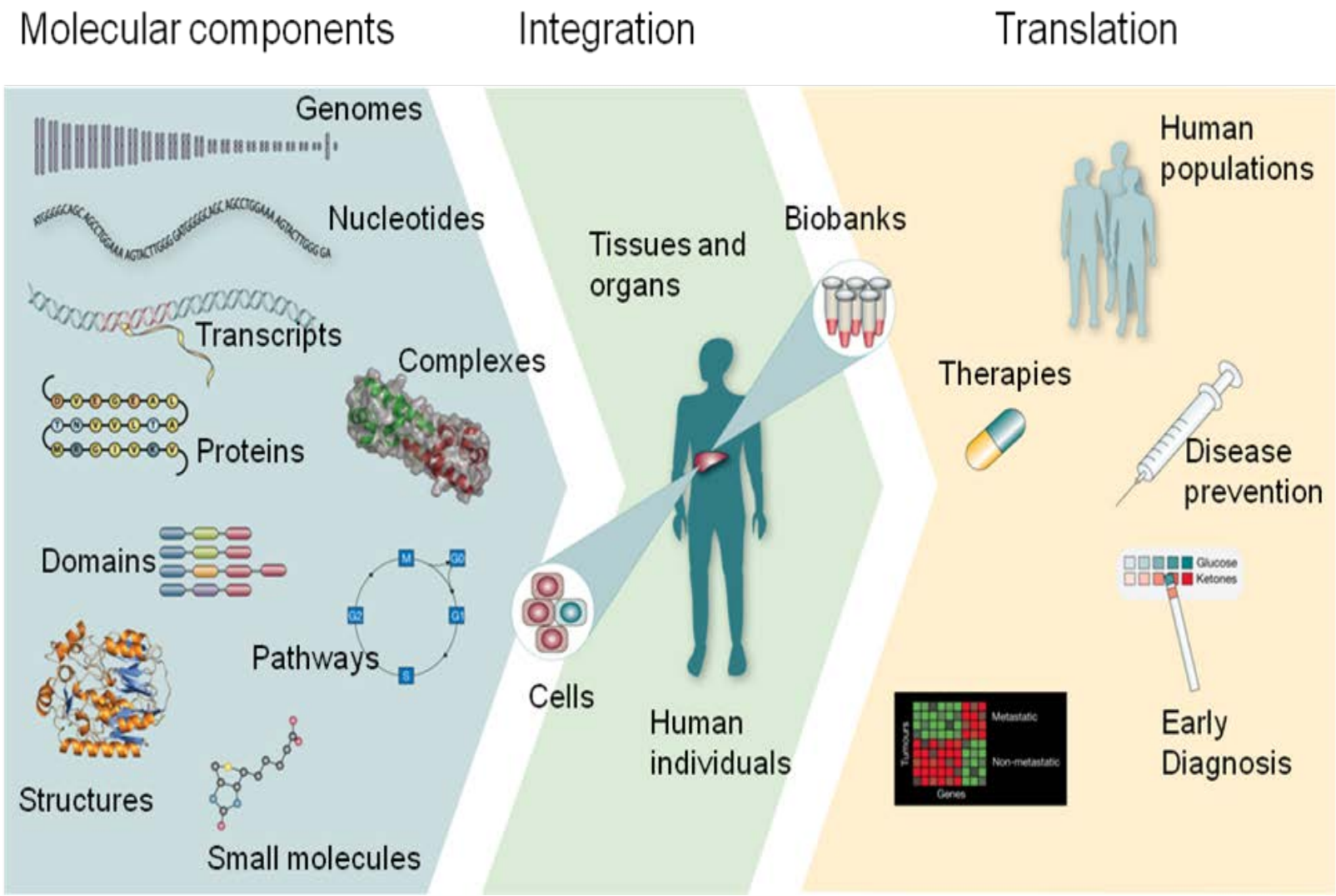
From molecules to phenotypes …
… from phenotypes to molecules



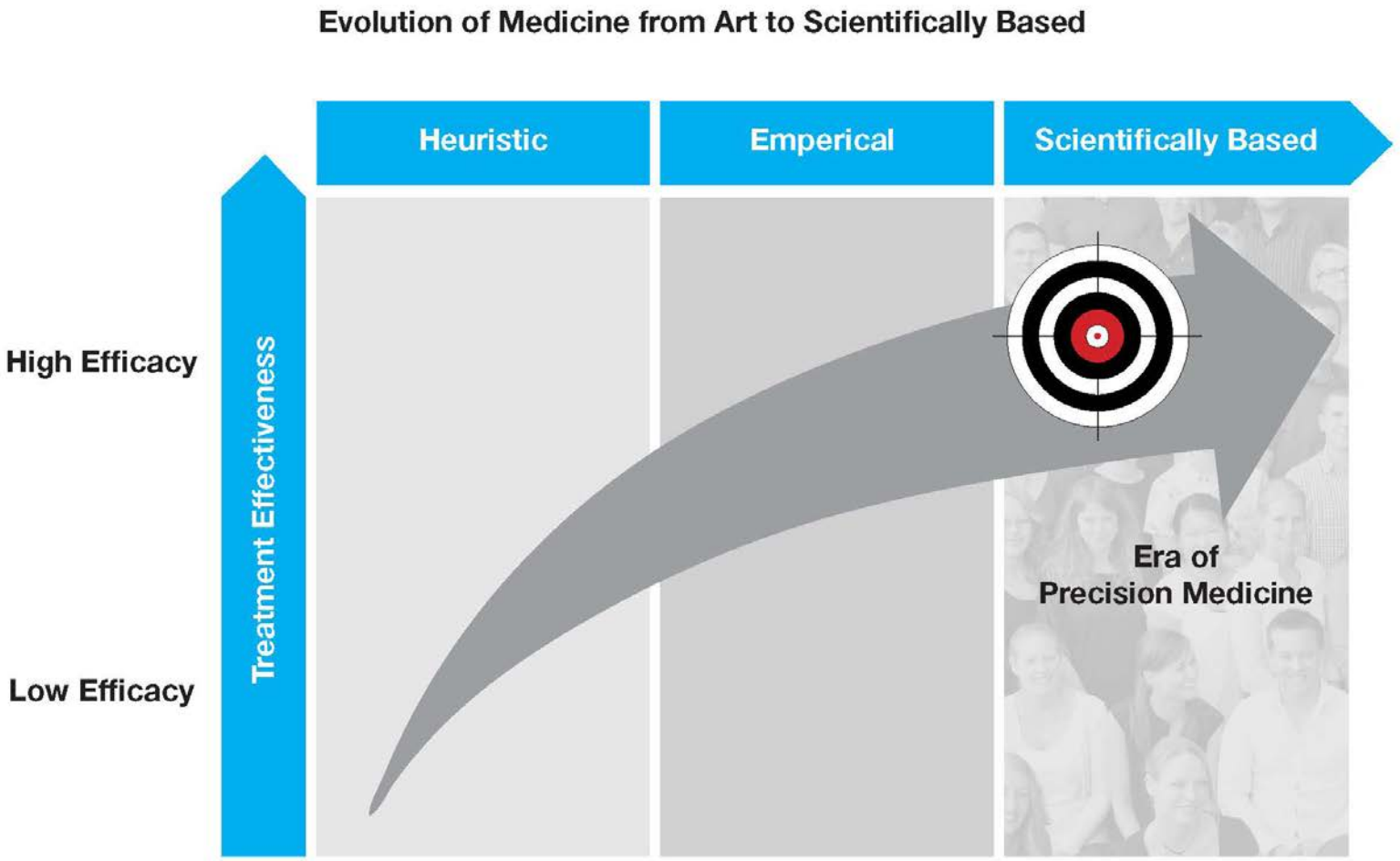
biomedbridges.eu

In 2001 we thought that sequence analysis was a solved problem and microarray analysis was where the action was. In 2012 it's the other way around.

Larry Hunter, 2012

Gibson TA (2012) The Roots of Bioinformatics
PLoS Comput Biol 8(8): e1002679.

Precision medicine



Fine-grained phenotype components

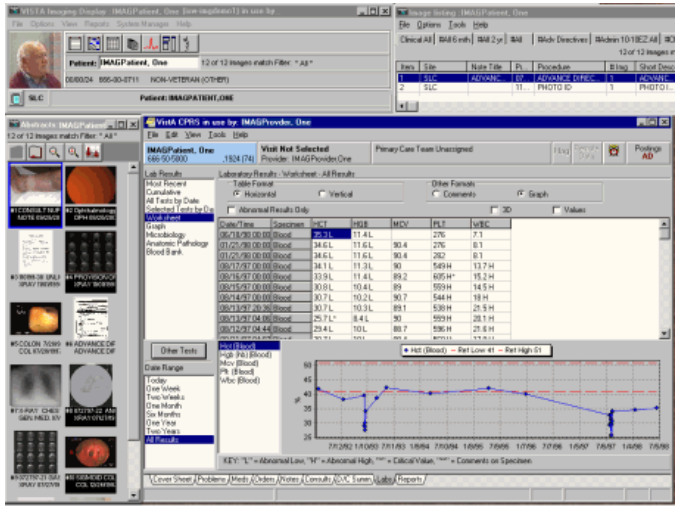
Beyond single disease analysis

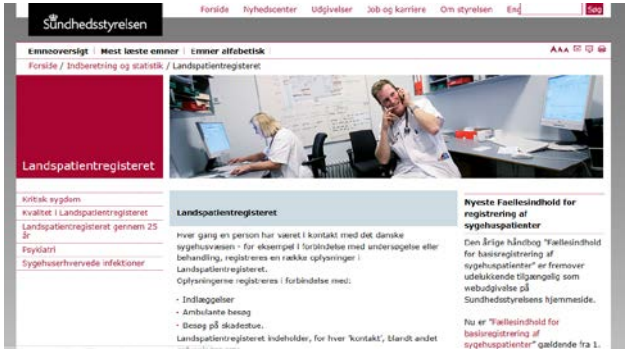
Disease-disease correlations

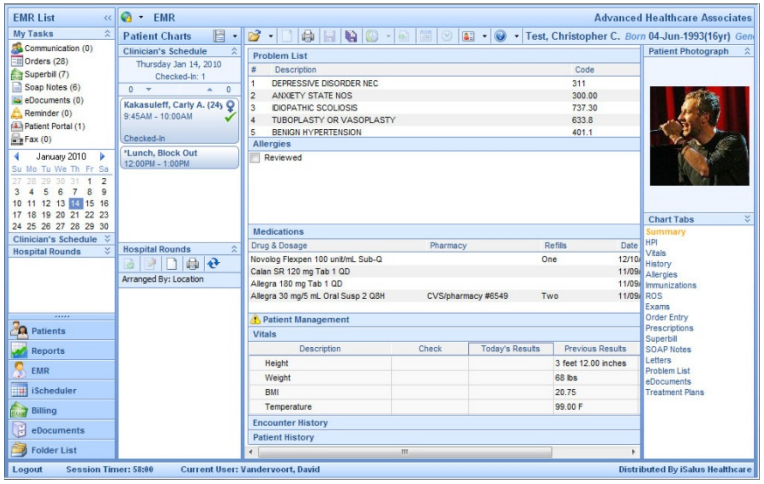
Disease-trajectories

What is potentially solely genetic and what is possibly treatment related?

Unstructured and structured data in electronic patient records, registries and questionaires

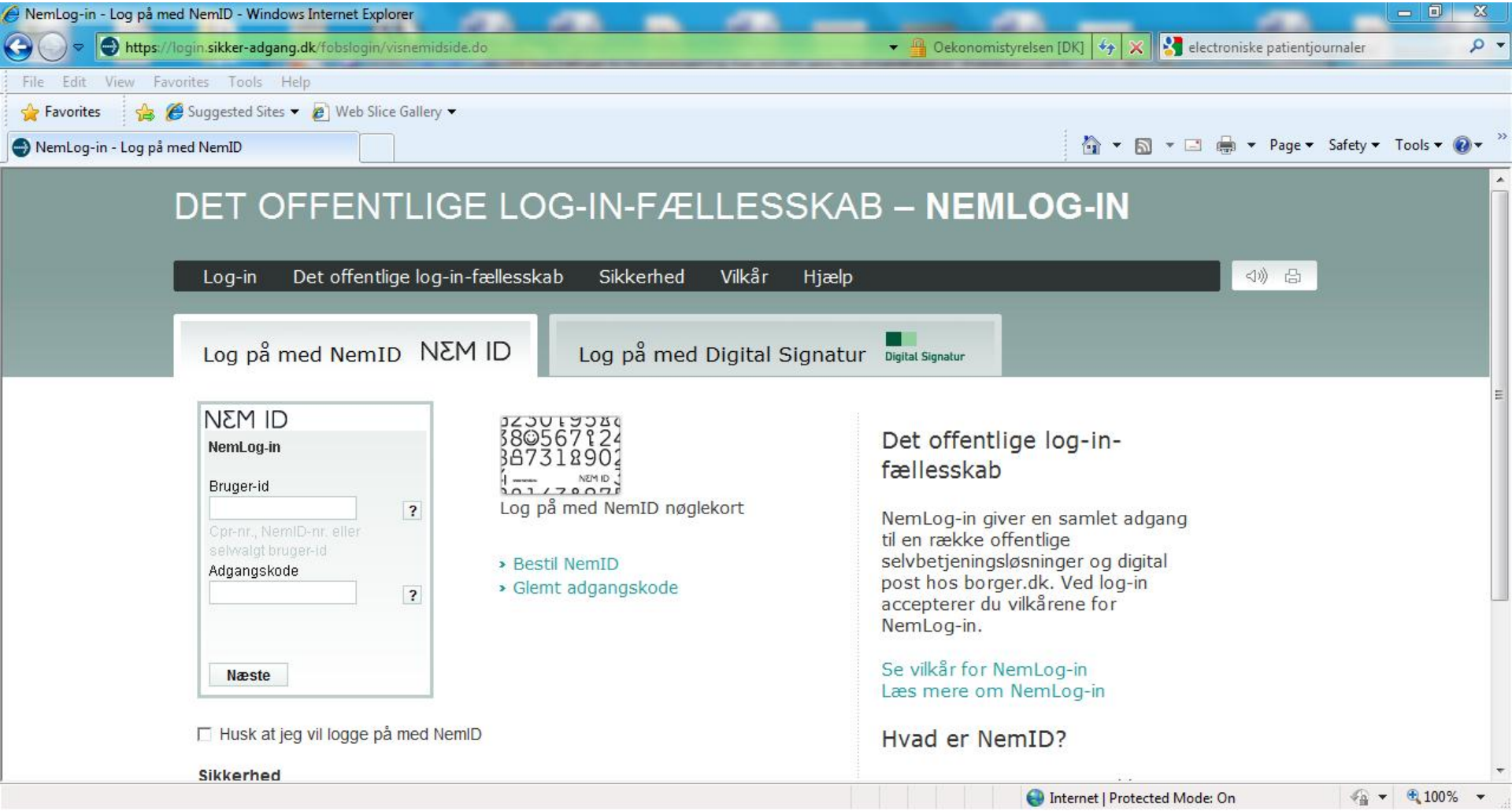






Login for all Danish citizens (> 15 years)

(Opt-out model)



Same secure login as for Home-banking, Tax, Social Services, Public dropbox etc. ect.

Personal identification number (Denmark)

From Wikipedia, the free encyclopedia

The Danish Personal Identification number (Danish: *CPR-nummer* or *personnummer*) is a national identification number, which is part of the personal information stored in the Civil Registration System (Danish: *Det Centrale Personregister*).

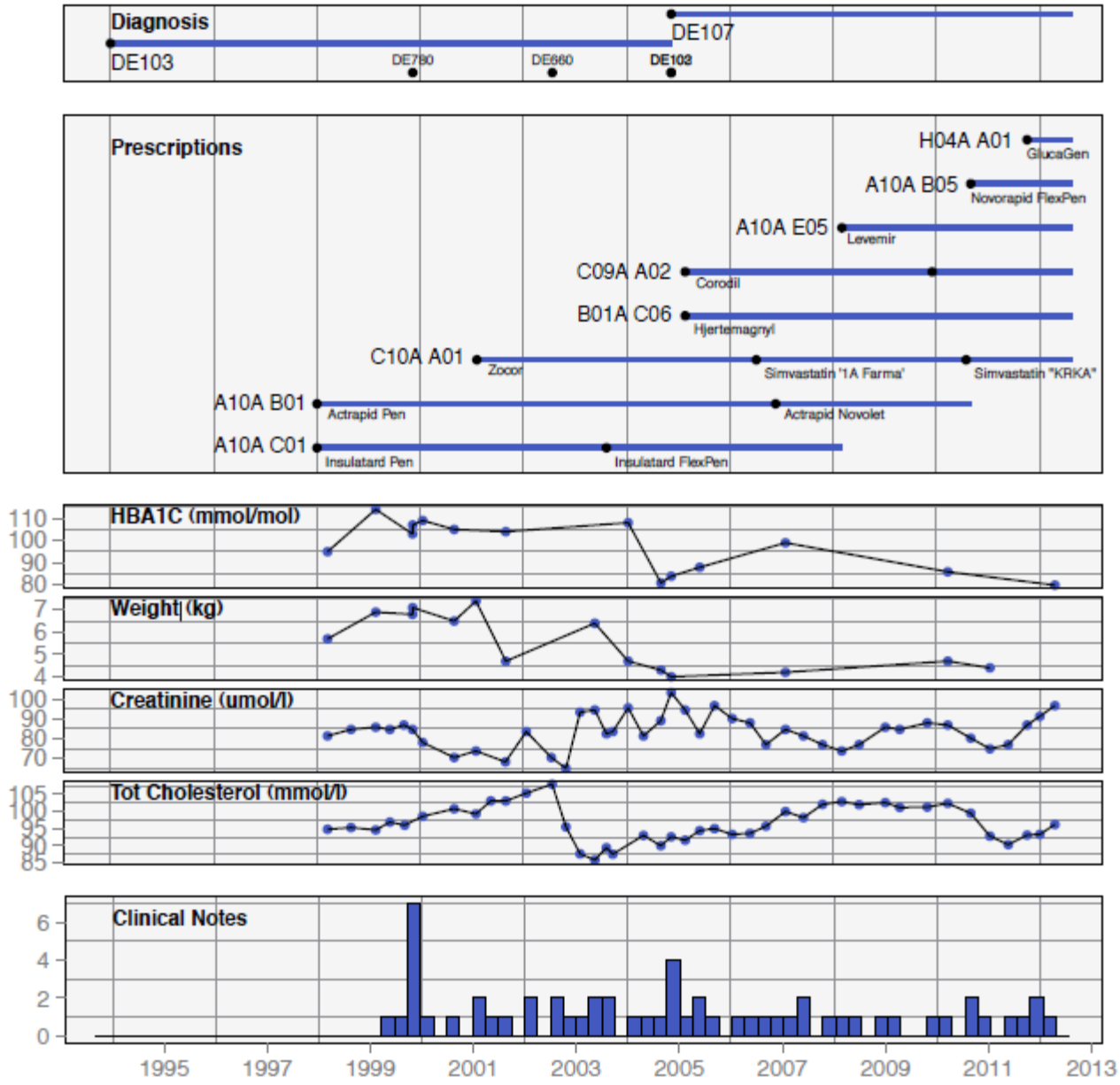
Was established in 1968.

It is a ten-digit number with the format DDMMYY-SSSS, where DDMMYY is the date of birth and SSSS is a sequence number. The first digit of the sequence number encodes the century of birth (so that centenarians are distinguished from infants), and the last digit of the sequence number is odd for males and even for females.

Any person registered as of 2 April 1968 or later in a Danish civil register, receives a personal identification number.

The civil register list only persons who:
- Are born in Denmark of a mother already registered in the civil register, *or*
- Have their birth or baptism registered in a 'Dansk Elektronisk Kirkebog (DNK)' (Danish electronic church-book), *or*
- Reside legally in Denmark for 3 months or more (non-Nordic citizens must also have a residence permit)

Danish citizens, including newborn babies, who are entitled to Danish citizenship, but are living abroad, do not receive a personal ID number, unless they move to Denmark.


Diagnosis
DE103
DE780
DE660
DE102
DE107
Prescriptions
H04A A01
GlucaGen
A10A B05
Novorapid FlexPen
A10A E05
Levemir
C09A A02
Corodil
B01A C06
Hjertemagnyl
C10A A01
Zocor
Simvastatin '1A Farma'
Simvastatin "KRKA"
A10A B01
Actrapid Pen
Actrapid Novolet
A10A C01
Insulatard Pen
Insulatard FlexPen
HBA1C (mmol/mol)
110
100
90
80
Weight (kg)
7
6
5
4
Creatinine (umol/l)
100
90
80
70
Tot Cholesterol (mmol/l)
105
100
95
90
85
Clinical Notes
6
4
2
0
1995
1997
1999
2001
2003
2005
2007
2009
2011
2013

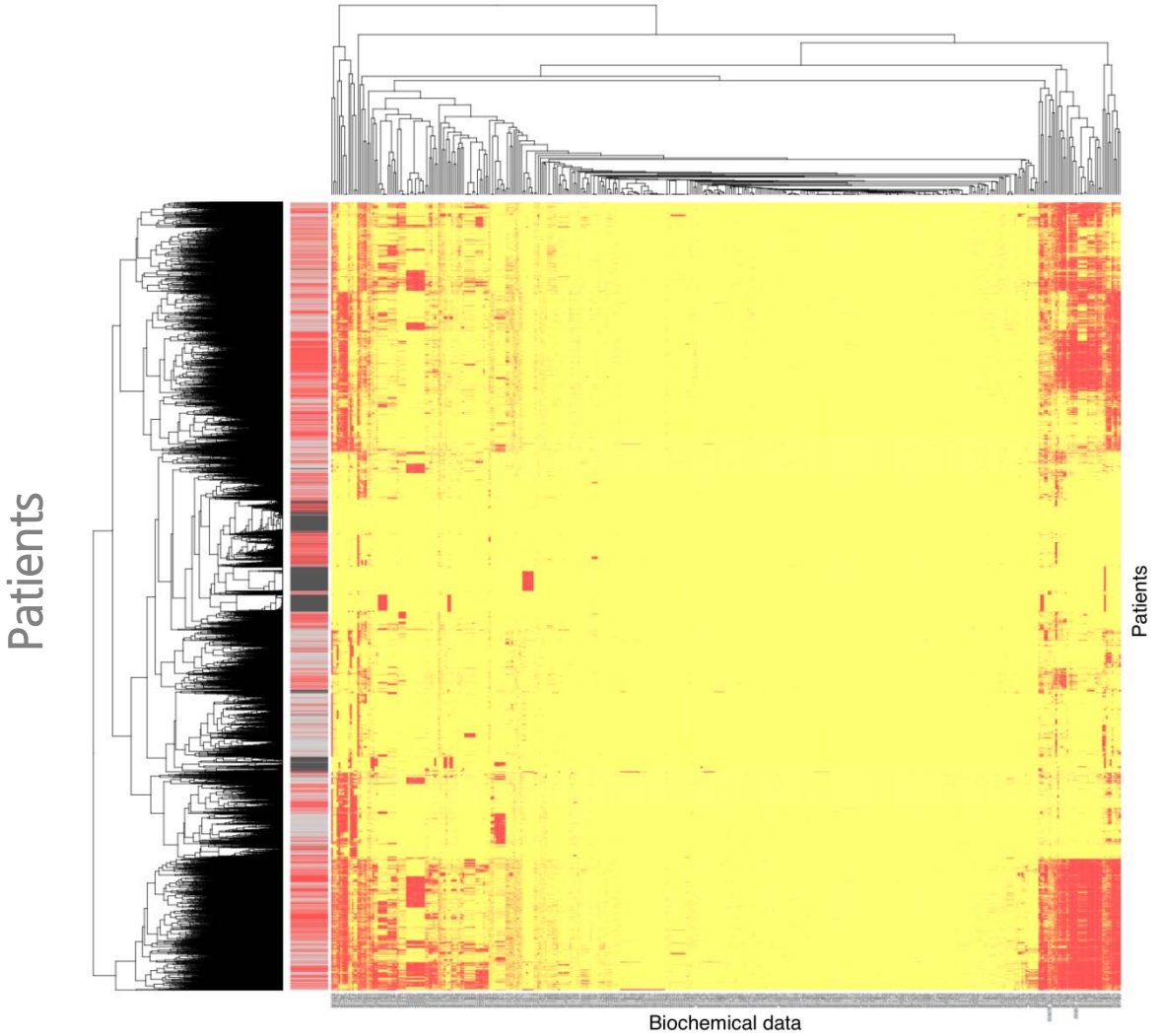
Wordfrequencies in Danish patient records

Testis cancer

Mental disorders

Infertility

Laboratory data from an entire diabetes clinic 12,000 patients



Lab data types 1:415

What is a precise phenotype?

Home » Harvard Health Blog » Overweight and healthy: the concept of metabolically healthy obesity

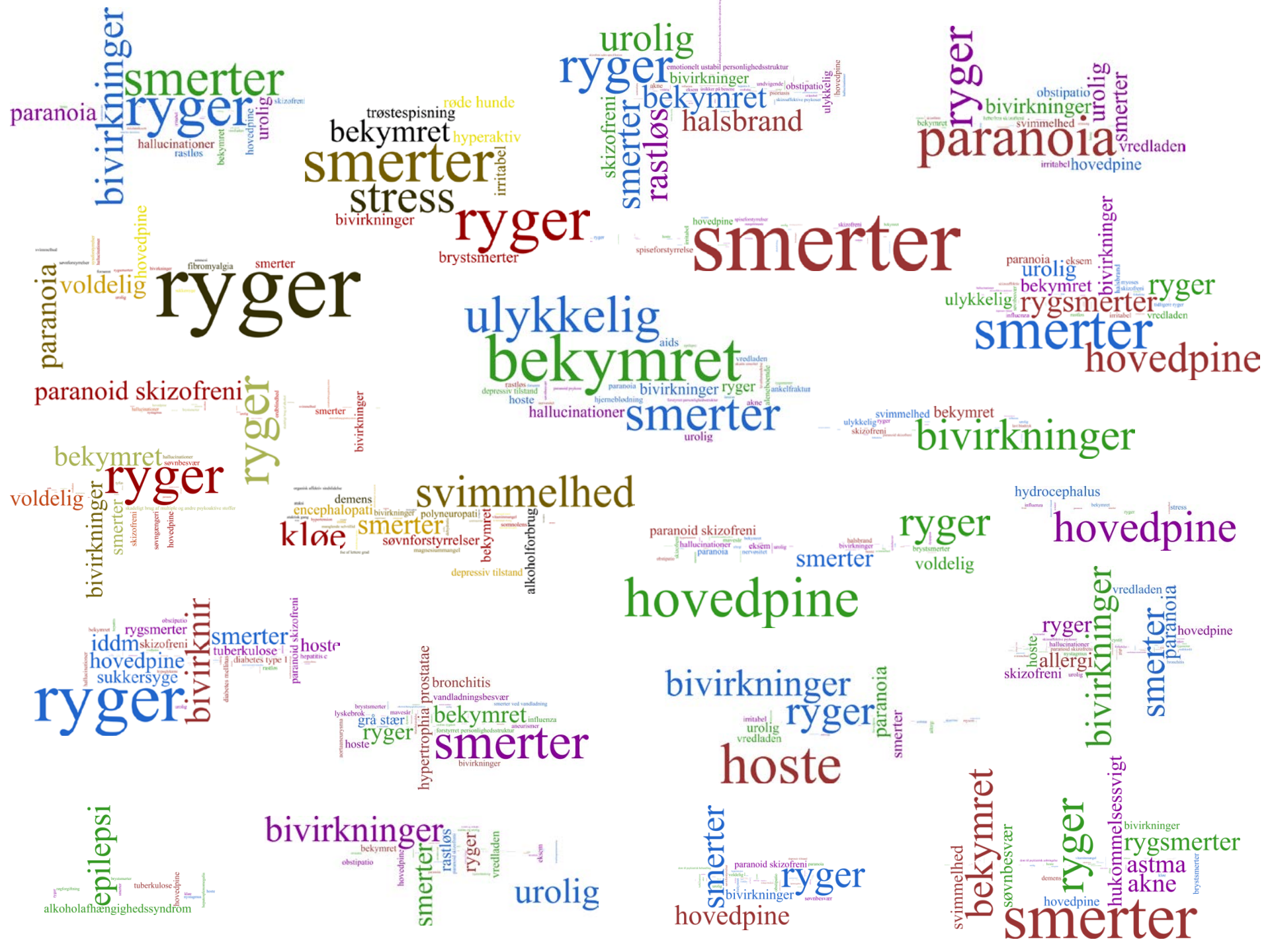
Overweight and healthy: the concept of metabolically healthy obesity

POSTED SEPTEMBER 24, 2013, 4:31 PM
Patrick J. Skerrett, Executive Editor, *Harvard Health*

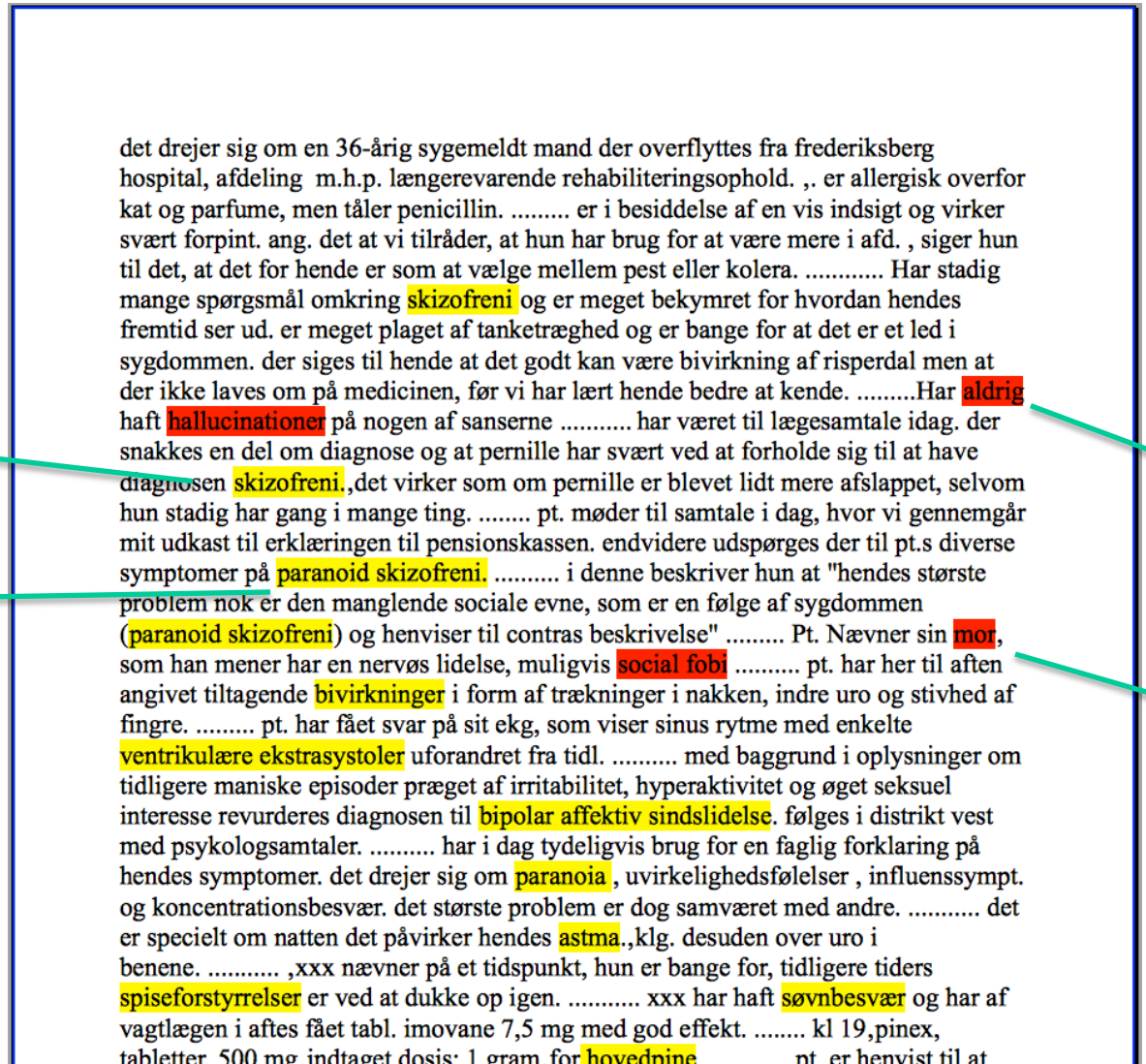
Carrying too many pounds is a solid signal of current or future health problems. But not for everyone. Some people who are overweight or obese mange to escape the usual hazards, at least temporarily. This weight subgroup has even earned its own moniker—metabolically healthy obesity.

Health professionals define overweight as a body-mass index (BMI) between 25.0 and 29.9, and obesity as a BMI of 30 or higher. (BMI is a measure of weight that takes height into consideration. You can **calculate your BMI here**.)

Most people who are overweight or obese show potentially unhealthy changes in metabolism. These include high blood pressure or high cholesterol, which damage


bivirkninger
smerter
ryger
paranoia
hallucinationer
rastløs
bekymret
hovedpine
urolig
skizofreni
trøstespisning
røde hunde
bekymret
hyperaktiv
smerter
irritabel
stress
bivirkninger
ryger
brystsmerter
urolig
ryger
bekymret
skizofreni
smerter
rastløs
halsbrand
obstipatio
bivirkninger
svimmelhed
paranoia
vredladen
irritabel
hovedpine
hovedpine
spiseforstyrrelse
smerter
paranoia
voldelig
hovedpine
fibromyalgia
smerter
ryger
paranoia
eksem
urolig
bekymret
bivirkninger
ulykkelig
rygsmerter
ryger
smerter
hovedpine
ulykkelig
aids
bekymret
vredladen
rastløs
depressiv tilstand
paranoia
bivirkninger
ryger
hoste
hjerneblødning
ankelfraktur
hallucinationer
smerter
urolig
paranoid skizofreni
smerter
bivirkninger
ryger
bekymret
hallucinationer
søvnbesvær
ryger
svimmelhed
bekymret
ulykkelig
skizofreni
bivirkninger
bekymret
ryger
voldelig
bivirkninger
smerter
skizofreni
søvngængeri
hovedpine
demens
encephalopati
bivirkninger
polyneuropati
svimmelhed
kløe
smerter
søvnforstyrrelser
bekymret
somnolens
depressiv tilstand
alkoholforbrug
paranoid skizofreni
hallucinationer
paranoia
eksem
nervøsitet
ryger
brystsmerter
smerter
voldelig
hydrocephalus
hovedpine
hovedpine
vredladen
bivirkninger
smerter
paranoia
hovedpine
ryger
hoste
allergi
skizofreni
rygsmerter
iddm
skizofreni
hovedpine
sukkersyge
tuberkulose
diabetes type 1
smerter
hoste
paranoid skizofreni
ryger
bivirknir
bronchitis
vandladningsbesvær
prostatae
hypertrophia
bekymret
influenza
grå stær
ryger
hoste
smerter
bivirkninger
bivirkninger
ryger
paranoia
smerter
irritabel
urolig
vredladen
hoste
epilepsi
alkoholafhængighedssyndrom
tuberkulose
hovedpine
bivirkninger
bekymret
obstipatio
hovedpine
smerter
rastløs
ryger
vredladen
eksem
urolig
smerter
paranoid skizofreni
ryger
bivirkninger
søvnbesvær
hovedpine
svimmelhed
bekymret
søvnbesvær
ryger
bivirkninger
rygsmerter
astma
akne
hukommelsessvigt
hovedpine
brystsmerter
smerter

Mine ICD10 dictionary terms from the medical record



det drejer sig om en 36-årig sygemeldt mand der overflyttes fra frederiksberg hospital, afdeling m.h.p. længerevarende rehabiliteringsophold. ,. er allergisk overfor kat og parfume, men tåler penicillin. ..... er i besiddelse af en vis indsigt og virker svært forpint. ang. det at vi tilråder, at hun har brug for at være mere i afd. , siger hun til det, at det for hende er som at vælge mellem pest eller kolera. ..... Har stadig mange spørgsmål omkring skizofreni og er meget bekymret for hvordan hendes fremtid ser ud. er meget plaget af tanketræghed og er bange for at det er et led i sygdommen. der siges til hende at det godt kan være bivirkning af risperdal men at der ikke laves om på medicinen, før vi har lært hende bedre at kende. .....Har aldrig haft hallucinationer på nogen af sanserne ..... har været til lægesamtale idag. der snakkes en del om diagnose og at pernille har svært ved at forholde sig til at have diagnosen skizofreni.,det virker som om pernille er blevet lidt mere afslappet, selvom hun stadig har gang i mange ting. ..... pt. møder til samtale i dag, hvor vi gennemgår mit udkast til erklæringen til pensionskassen. endvidere udspørges der til pt.s diverse symptomer på paranoid skizofreni. ..... i denne beskriver hun at "hendes største problem nok er den manglende sociale evne, som er en følge af sygdommen (paranoid skizofreni) og henviser til contras beskrivelse" ..... Pt. Nævner sin mor, som han mener har en nervøs lidelse, muligvis social fobi ..... pt. har her til aften angivet tiltagende bivirkninger i form af trækninger i nakken, indre uro og stivhed af fingre. ..... pt. har fået svar på sit ekg, som viser sinus rytme med enkelte ventrikulære ekstrasystoler uforandret fra tidl. ..... med baggrund i oplysninger om tidligere maniske episoder præget af irritabilitet, hyperaktivitet og øget seksuel interesse revurderes diagnosen til bipolar affektiv sindslidelse. følges i distrikt vest med psykologsamtaler. ..... har i dag tydeligvis brug for en faglig forklaring på hendes symptomer. det drejer sig om paranoia , uvirkelighedsfølelser , influenssympt. og koncentrationsbesvær. det største problem er dog samværet med andre. ..... det er specielt om natten det påvirker hendes astma.,klg. desuden over uro i benene. ..... ,xxx nævner på et tidspunkt, hun er bange for, tidligere tiders spiseforstyrrelser er ved at dukke op igen. ..... xxx har haft søvnbesvær og har af vagtlægen i aftes fået tabl. imovane 7,5 mg med god effekt. ..... kl 19,pinex, tabletter, 500 mg, indtaget dosis: 1 gram, for hovedpine. pt. er henvist til at

F20

F200

Negation

Family

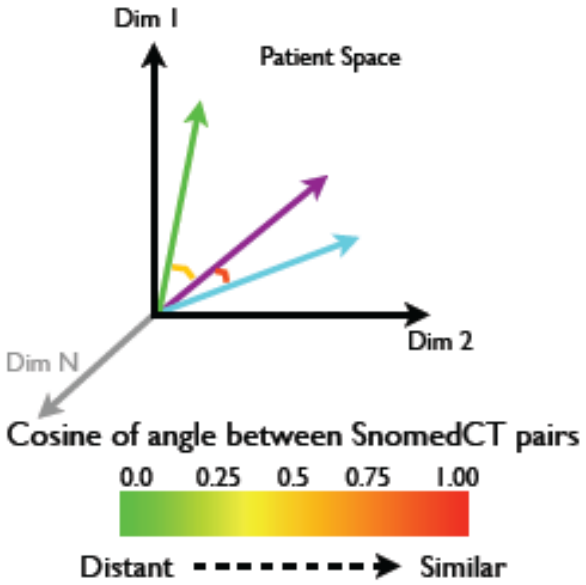
Compare patients by ICD10 terms mined and assigned in terminology space

Patient 1

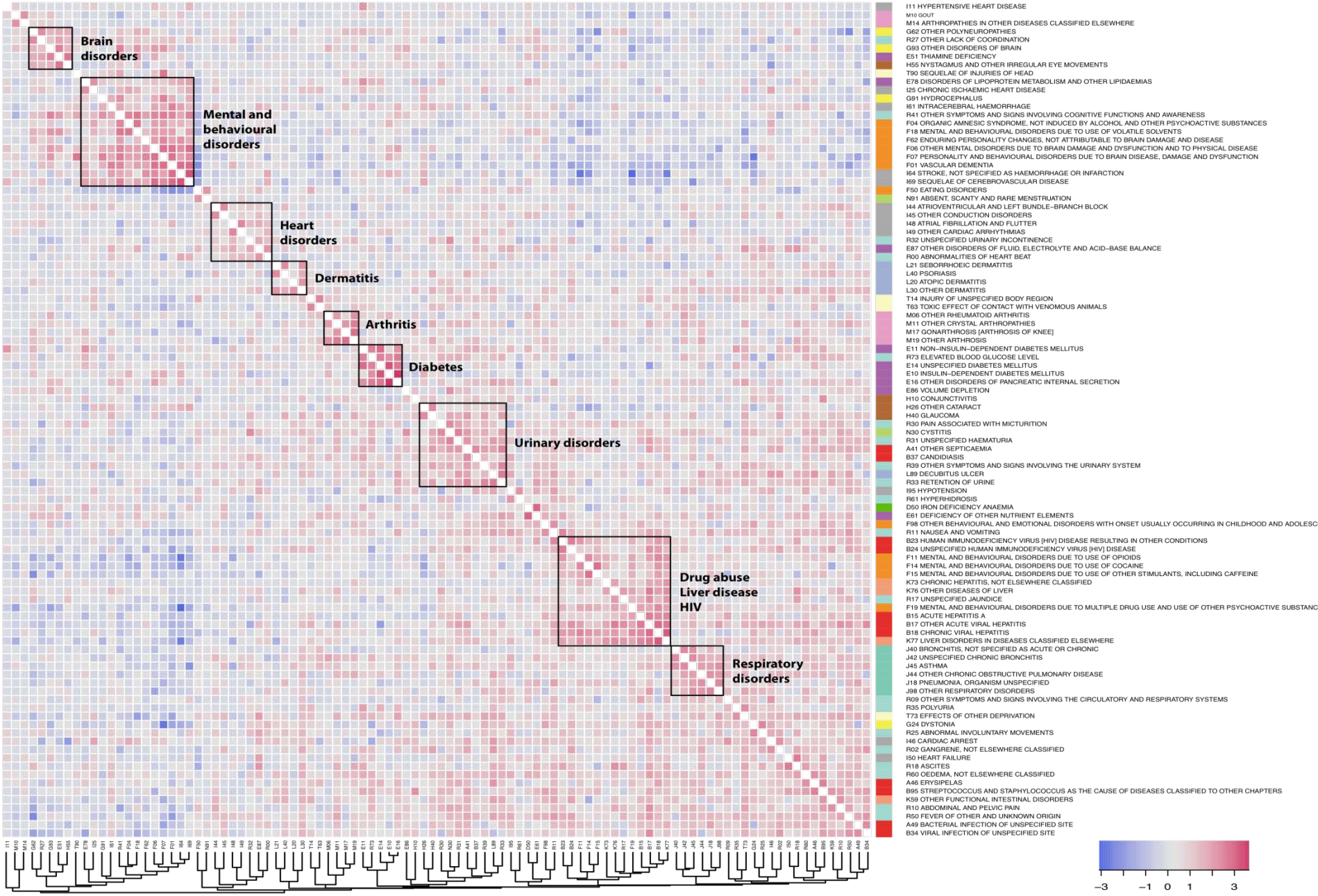
Patient 2

Patient 3

PID	248153007	88311004	386633000	...
pat1	1	0	0	
pat2	0	1	0	
pat3	0	0	1	
...				



Roque et al. PLoS Comp. Biol. 2011,
Jensen et al., Nature Rev. Genet. 2012



Roque et al. PLoS Comp. Biol. 2011,
Jensen et al., Nature Rev. Genet. 2012

Significant comorbidities among complex and Mendelian disorders (110M patients, registry data)

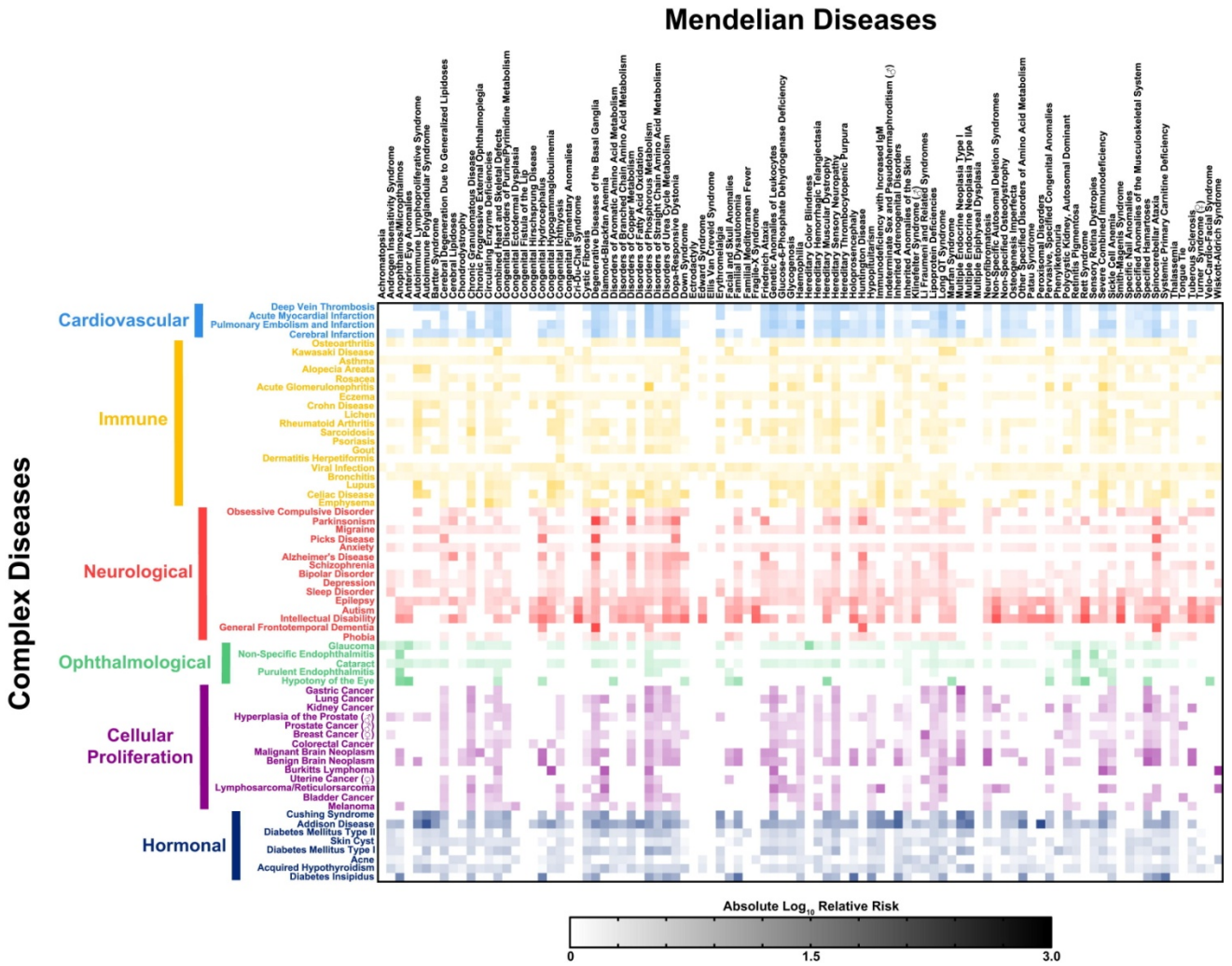


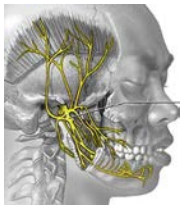
Table 1. The Clinical Record Data Sets Utilized in This Study

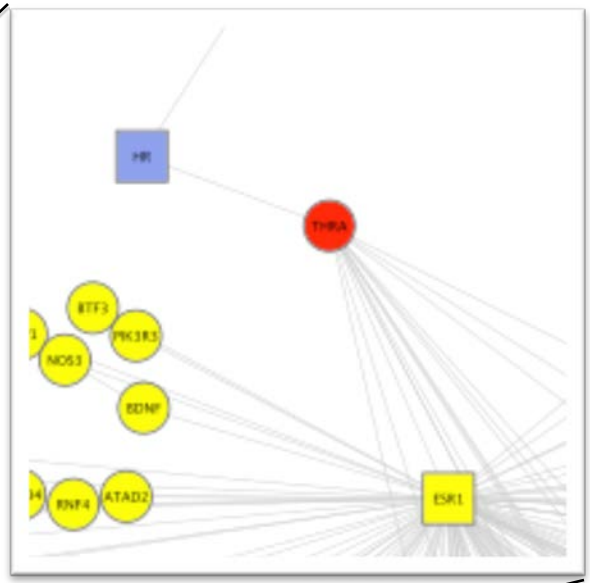
Data Set	Description	Encoding Type	Number of Unique Patients
CU	Columbia University, 1985–2003	ICD9	1,505,822
DK	Denmark; database covering most of the country's population	ICD10	6,214,312
NYPH	New York Presbyterian Hospital and Columbia University; 2004–present	ICD9	767,978
SU	Stanford University	ICD9	806,369
TX	University of Texas at Houston	ICD9	1,599,528
UC	University of Chicago	ICD9	146,989
USA	MarketScan insurance claims data set	ICD9	99,143,849
MED	Medicare database	ICD9	13,039,018
	Total:		123,223,865

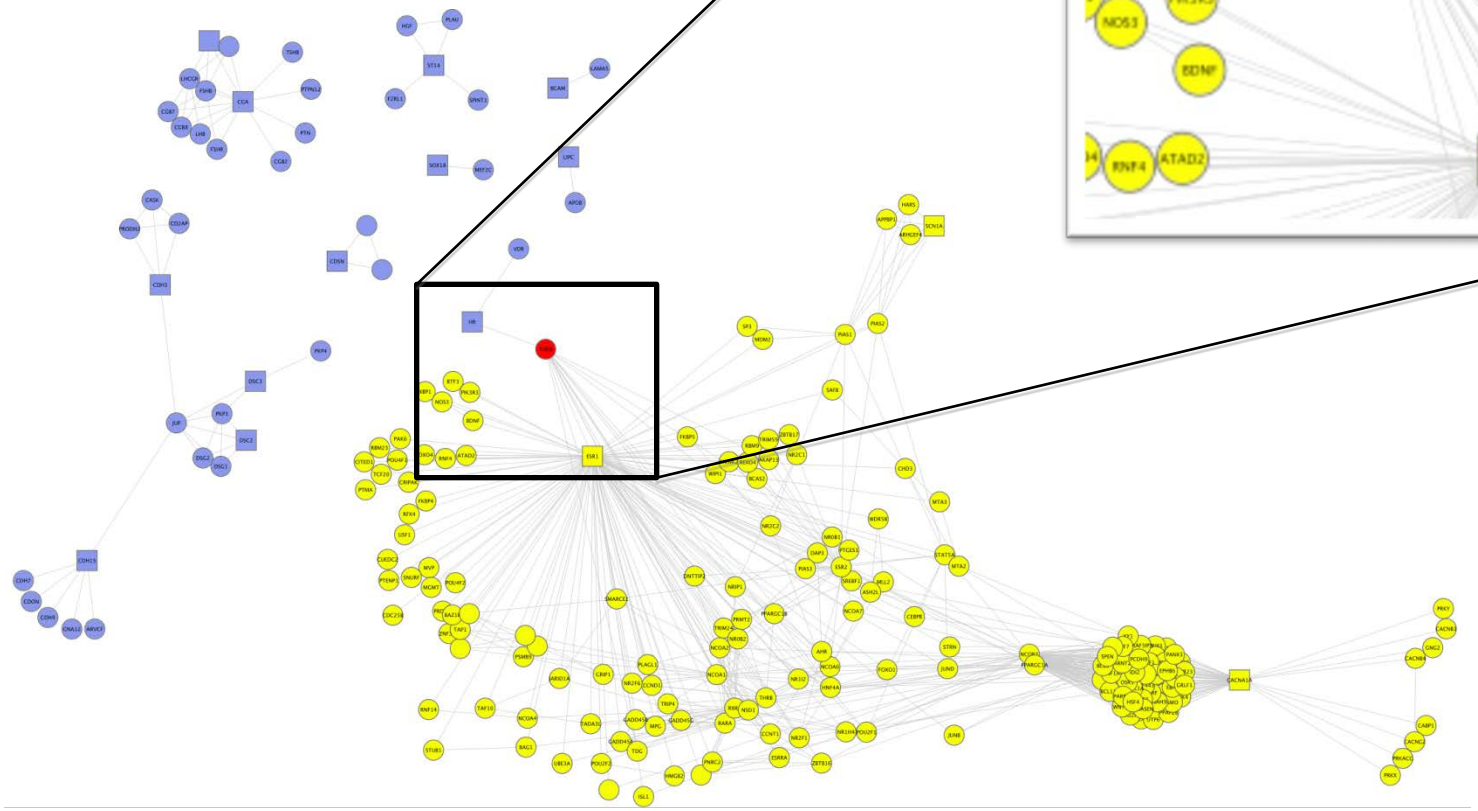
This table provides a brief description, the ICD encoding type, and the size of each data set. The MED data set was used for comparison and was not included in the full meta-analysis.

Blair et al., Cell 2013

Alopecia L65 and Migraine G43







The shared gene is THRA, connected to ESR1 from migraine (yellow) and HR from Alopecia (blue). Seed genes are squares, red is the connection gene. The HR-THRA PPI has a score of 1 in the scored interactome.

Language independence and interoperability of ICD10 and SNOMED CT terms

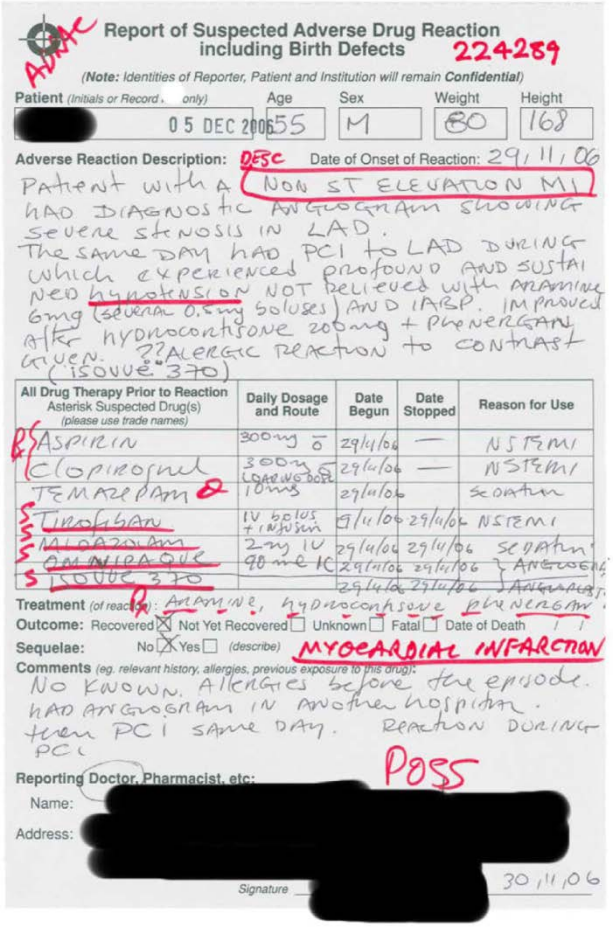
mand	male
orkiektomi	orchiectomy
tobak	tobacco
biopsi	biopsy
ingentegn	absence of signs
cyanose	cyanosis
splenomegali	splenomegaly
lilletestikel	small testicle
smerter	pain
stimulationstest	stimulation test
testosteron	testosterone
protein	protein
palpation	palpation
injektion	injection
hormon	hormone
volumen	loudness
hormonbehandling	hormone therapy
ordination	prescription
svedeture	hot sweats
ingenabnormitetfundet	no abnormality detected
ingenklager	no complaints
stor tumor	large tumor
anbefalingom	recommendation to
pet-scanning	positron emission tomography
kemoterapi	chemotherapy
cisplatin	cisplatin
bleomycin	bleomycin
dybvenetrombose	deep venous thrombosis
venetrombose	venous thrombosis
ryger	smoker
bivirkninger	adverse reaction
fjernelse	removal
gemcitabin	gemcitabine
obstipation	obstipation
halsbrand	waterbrash
krise	crisis
vedligeholdelsesbehandling	maintenance therapy
blod	blood
ifosfamid	ifosfamide
chrom	chromium
begravelse	burial
mesna	mesna
kvalmeogopkastning	nausea and vomiting
alkohol	alcohol
krampe	cramp
magnesium	magnesium
methotrexat	methotrexate
vinblastin	vinblastine
carboplatin	carboplatin
oxaliplatin	oxaliplatin
hoste	cough
krepitation	crepitation
krampeanfald	seizure
opkast	vomitus
interstitiellungesygdom	interstitial lung disease
lungesygdom	disorder of lung
pindolol	pindolol
etoposid	etoposide
d-dimer	d-dimer
spredning	dispersion
ct afthorax	computed tomography of chest
ct-scanning af thorax og abdomen	computed tomography of chest and abdomen
hovedpine	headache
gastrit	gastritis
pankreatit	pancreatitis
narkose	narcosis
incision	incision
diurese	diuresis
kalium	potassium
drikker	does drink
mobilisering	mobilization
lagvislukning	layer closure procedure
lukning	reparative closure
god prognose	prognosis good
pethidin	meperidine
feber	fever
ekspektorat	sputum
icterus	jaundice
hypogonadisme	hypogonadism
gift	married
flebografi	venography
hjertebanken	pounding heart
struma	goiter
hepatosplenomegali	hepatosplenomegaly
information	informing
hepatomegali	large liver
brystsmerter	chest pain
stabilisering	stabilization
muskelsmerter	muscle pain
pneumoni	pneumonia
ledsmerter	joint pain

提要該当ﾍﾟｰｼﾞ	旧（ＩＣＤ－１０　準拠）	新（ＩＣＤ－１０　２００３年版　準拠）
	痴呆の記載箇所	認知症に変更
217	下記の４桁分類項目は項目F10 - F19に使用する **.0　急性中毒**	下記の４桁分類項目は項目F10 - F19に使用する。 **.0　急性中毒** **除外**：薬物による中毒<poisoning>を意味する場合(T36-T50)
221	**精神分裂病，分裂病型障害および妄想性障害（F20-F29）** 精神分裂病または分裂病の記載箇所	**統合失調症，統合失調症型障害および妄想性障害（F20-F29）** 統合失調症に変更
221	**F20　精神分裂病** 精神分裂病または分裂病の記載箇所	**F20　統合失調症** 統合失調症に変更
222	**F20.0　妄想型分裂病** 精神分裂病または分裂病の記載箇所	**F20.0　妄想型統合失調症** 統合失調症に変更
222	**F20.1　破瓜型分裂病** 精神分裂病または分裂病の記載箇所	**F20.1　破瓜型統合失調症** 統合失調症に変更
222	**F20.2　緊張型分裂病** 精神分裂病または分裂病の記載箇所	**F20.2　緊張型統合失調症** 統合失調症に変更
223	**F20.3　型分類困難な分裂病** 精神分裂病または分裂病の記載箇所	**F20.3　型分類困難な統合失調症** 統合失調症に変更
223	**F20.4　分裂病後抑うつ**	**F20.4　統合失調症後抑うつ**

Which disease-disease and symptom correlations are treatment related?

Spontaneous reports

- **Heavily trusted**
- **Underreporting and biases**
- **Data quality issues**



Report of Suspected Adverse Drug Reaction including Birth Defects 224289

(Note: Identities of Reporter, Patient and Institution will remain Confidential)

Patient (Initials or Record only) [redacted] 05 DEC 2006 | Age 55 | Sex M | Weight 80 | Height 168

Adverse Reaction Description: DESC Patient with a NON ST ELEVATION MI had diagnostic angiogram showing severe stenosis in LAD. The same day had PCI to LAD during which experienced profound and sustained hypotension not believed with aramine 6mg (several 0.5mg boluses) and IABP. Improved after hydrocortisone 200mg + phenergan given. ??Allergic reaction to contrast (isovue 370)

Date of Onset of Reaction: 29/11/06

All Drug Therapy Prior to Reaction Asterisk Suspected Drug(s) (please use trade names)	Daily Dosage and Route	Date Begun	Date Stopped	Reason for Use
PS ASPIRIN	300mg O	29/11/06	—	NSTEMI
clopidogrel	300mg LOADING DOSE	29/11/06	—	NSTEMI
TEMAZEPAM O	10mg	29/11/06		SEDATION
S tirofiban	IV BOLUS + INFUSION	29/11/06	29/11/06	NSTEMI
S MIDAZOLAM	2mg IV	29/11/06	29/11/06	SEDATION
S OMNIPAQUE	90 ml IC	29/11/06	29/11/06	ANGIOGRAM
S ISOVUE 370		29/11/06	29/11/06	ANGIOGRAM

Treatment (of reaction): aramine, hydrocortisone phenergan

Outcome: Recovered ☒ Not Yet Recovered ☐ Unknown ☐ Fatal ☐ Date of Death / /

Sequelae: No ☒ Yes ☐ (describe) MYOCARDIAL INFARCTION

Comments (eg. relevant history, allergies, previous exposure to this drug): No known allergies before the episode. had angiogram in another hospital. then PCI same day. Reaction during PCI

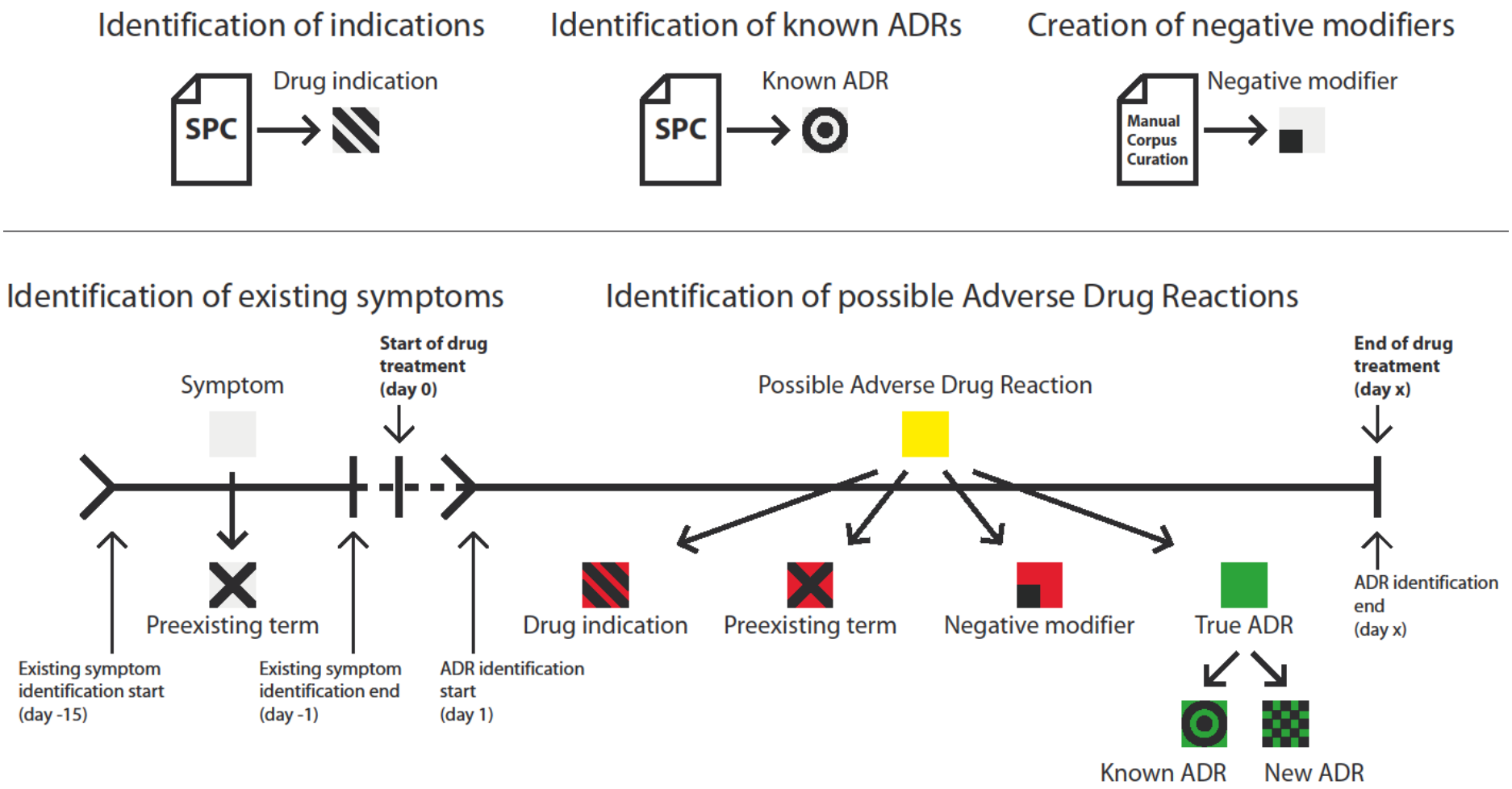
Reporting Doctor, Pharmacist, etc: POSS

Name: [redacted]

Address: [redacted]

Signature [redacted] 30/11/06

Text mining Adverse Drug Reactions
(using 7,500 drug names and 21,000 ADRs)



Eriksson et al. Drug Safety 2014

Text mining of drug names, ADE/ADRs, diagnoses, …

Removed ADR - no corresponding structured data

Behandlet med Zyprexa 5 mg fra 3. til 24.6.99 og 10 mg fra 24. til 29.6.99 med nogen effekt på tankeforstyrrelser, men seponeret pga appetitøgning. Herefter Risperdal 2 mg stigende til 4 mg i perioden 29.6. til 12.7.99, men seponeret på grund af uro i kroppen og "ostek-lokkefornemmelse". Herefter Orap 2 mg fra 2.8. stigende til 3 mg fra 30.8.99 med god effekt på tankeekko og tankemylder. Behandlet med Zoloft 50 mg fra maj 98 til maj 99 med noget virkning på depressive symptomer, men seponeret på grund af natlig svedtendens. Siden 14.7.99 Efexor 75 mg med nogen effekt på antallet og sværhedsgraden af kortvarige depressive episoder

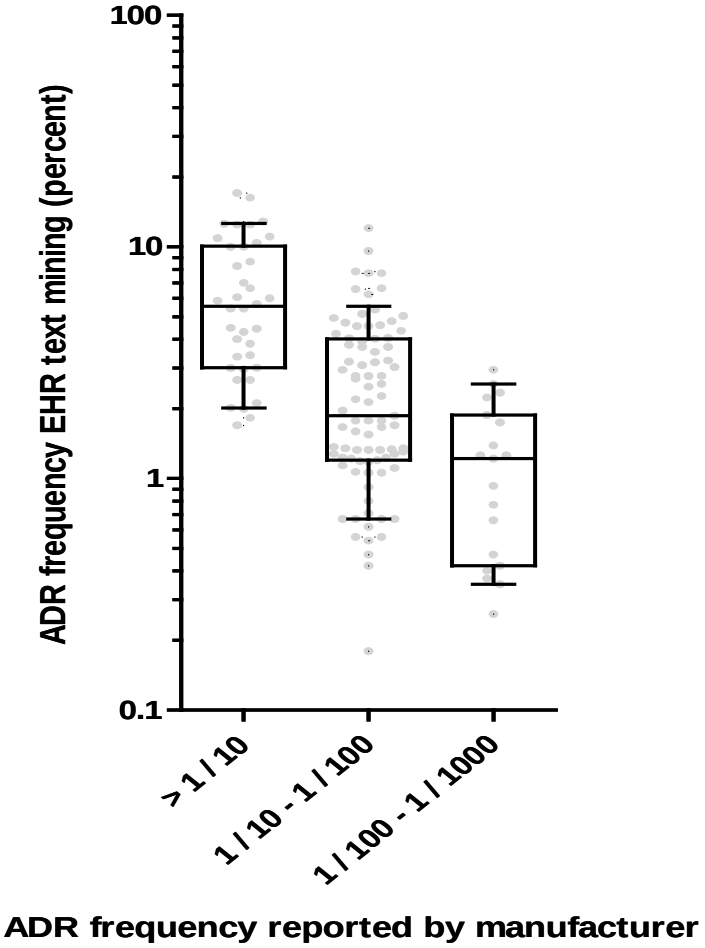
se venligst under allergier. Desuden forsøgt beh med Zyprexa. sep grundet vægtøgning, træthed og manglende effekt. Risperdal ord med nogen effekt tillagt dogmatil(1999). Efterfølgende aurorix beh seponeret i 1999. Startede istedet remeron. Aktuel medicindois, jvf ud-skrivningsnotat fra U12 samt EPJmedicinliste.

tbl. leponex 100+0+0+200 mg,tbl. rivotril 0,5 +0+1+ 0 mg,tbl. arintapin 0+0+0+30 mg,tbl. klomipramin 0+0+0+25 mg,tbl. imoclone 7,5 mg nocte,tbl. rivotril 2 mg p.n. max x 1 dgl. tbl. marevan a 2,5 mg efter skema,tbl. magnesia 1 g p.n. laxoberaldr. 7,5 mg /ml 15 dr. p.n. mix. link 150 mg /ml 15 ml p.n. max x 3 dgl. Figensaft 20+0+20+0 ml,Pt. er aktuelt,CAVE,tricykliske antidepressiva. Dette kan dog ikke bekræftes og pt. har tidl. fået imipramin, som han har tålt godt, hvorfor der er ansøgt om ophævelser af denne cave på højere niveau. Har tidl. fået zyprexa som blev sep. grundet vægtøgning, træthed og manglende effekt

Removed ADR - negation and subject identification

...Jeg mener fortsat, at han har brug for medicin, da han i går fx var meget vred og følte sig utryg og angst og har haft svært ved at sove. Dette synes pt. at accepterer. Jeg tilbyder herefter Zyprexa i stedet for Risperdal, pt. afviser dette, da han ved medpatient har fået denne medicin og har fået øget appetit, dette vil han ikke. Har ikke tidligere fået antipsykotisk medicin. Accepterer herefter Cisordinol, startende på en lille dosis. Accepterer også angstdæmpende medicin i dagtiden. Angiver, at når han bliver vred kan han godt styre det. Synes det hjalp i går noget at få Nozinan. Virker fortsat garderet. Siger intet uopfordret. Sparsomt sygdomsindsigt. Er ikke vredladen. Er i dag er heller latent aggressiv...

Frequency of Adverse Drug Reactions from text mining



Comparison between the 150 most statistically significant extracted ADR frequencies and the stated frequencies in the SPC by the manufacturer. Dots are showing the individual extracted frequencies.

Out of the 150 are 6 left out, according to the SPC these are having a frequency < 1/1000 and the corpus is not large enough to detect these with a satisfying frequency.

Recall of 75,1% and a precision of 95,0%. (manual curation of 200 records)

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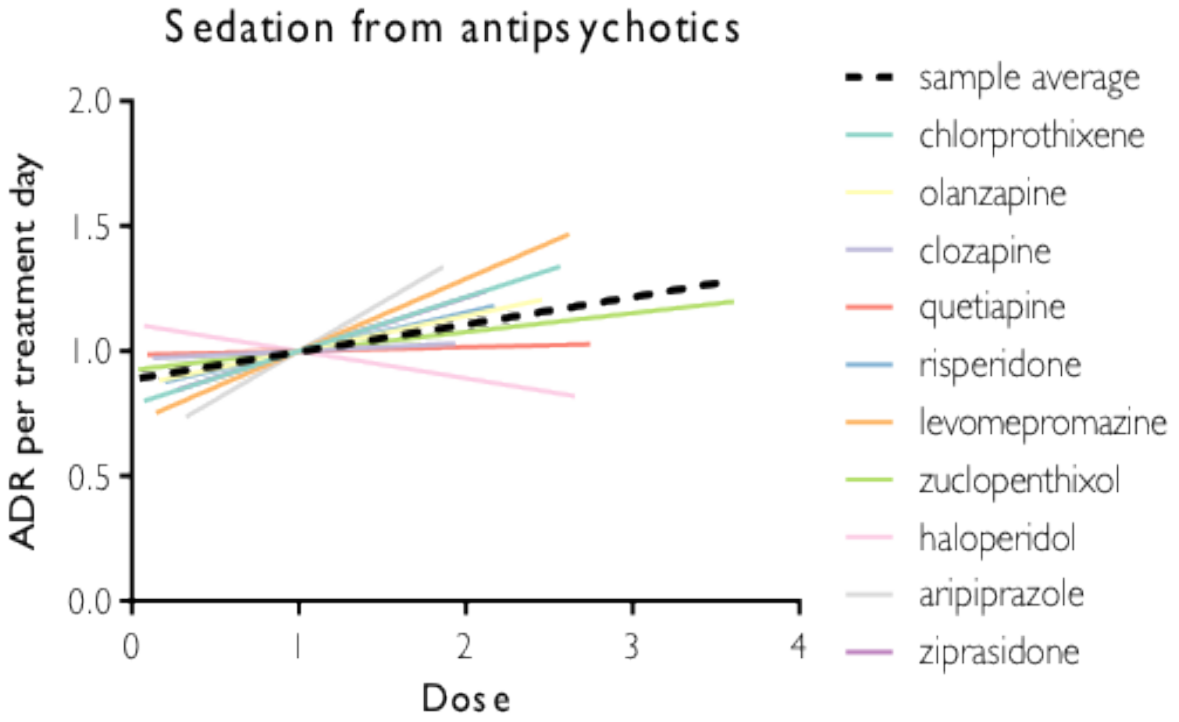
ADR-dose dependencies

Dosages from structured medication data

Sedation is the most occurring ADR in the corpus

Drugs selected are 10 antipsycotics (a class known to cause sedation)



ADRs and doses are normalized on multiples of the minimum dose prescribed of each drug.

Plot for 21 days steady dosage data is visualized, sample average slope 0.1105 (95% CI, 0.03085-0.1901), non-zero slope p-value was 0,0074, all individual drug slopes are positive except for haloperidol.

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Possible ADRs?

Drug substance	ADE	p-value
Dipyridamole	Visual impairment	4.375e-04
Simvastatin	Personality changes	8.408e-08
Citalopram	Psychosis	8.807e-04
Bendroflumethiazide	Apoplexy	8.46e-03
Chlordiazepoxide	Nystagmus	4.03e-08

X1	Y1	p1
X2	Y2	p2
....		

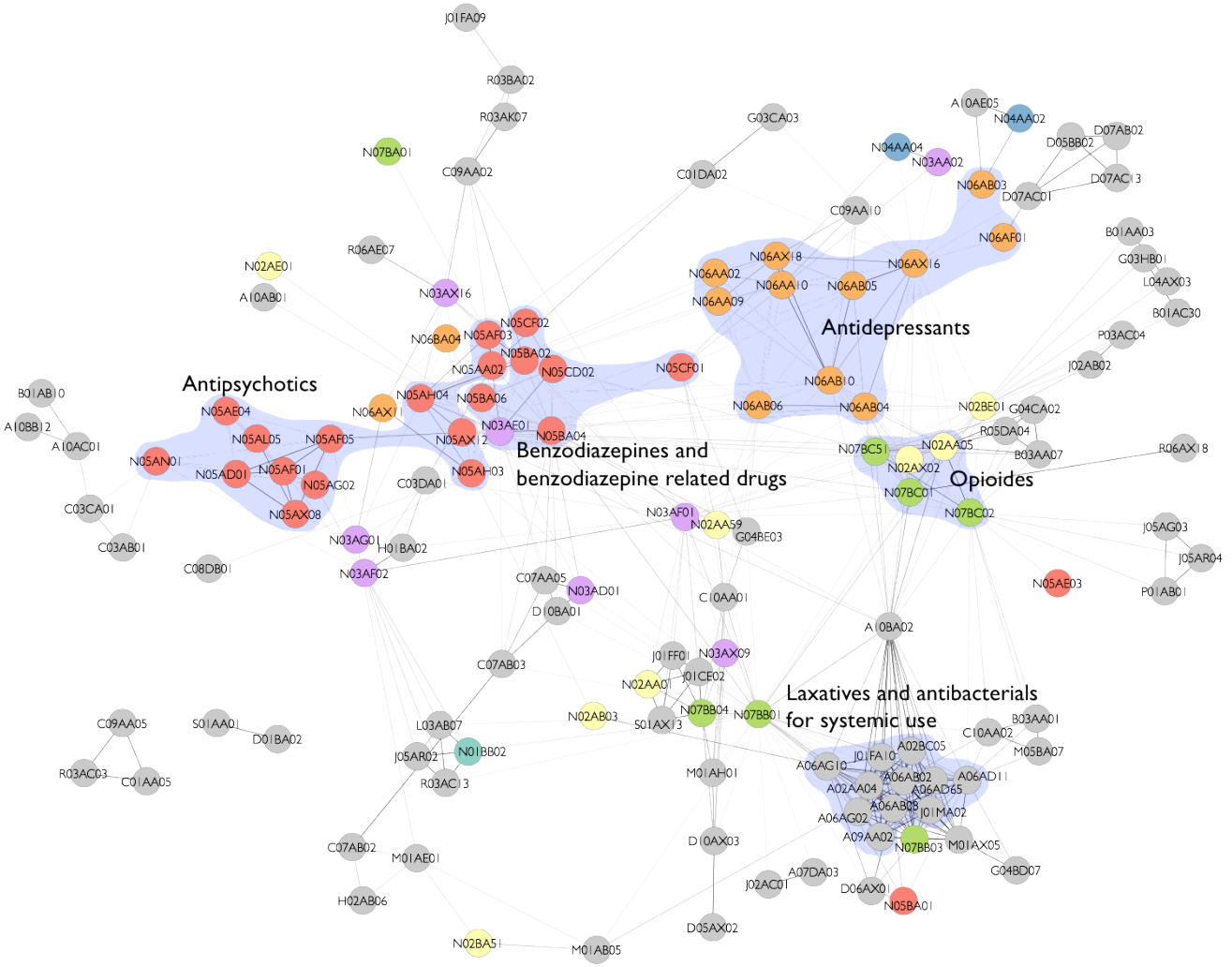
p-values are multiple testing corrected

Drug-ADR similarities

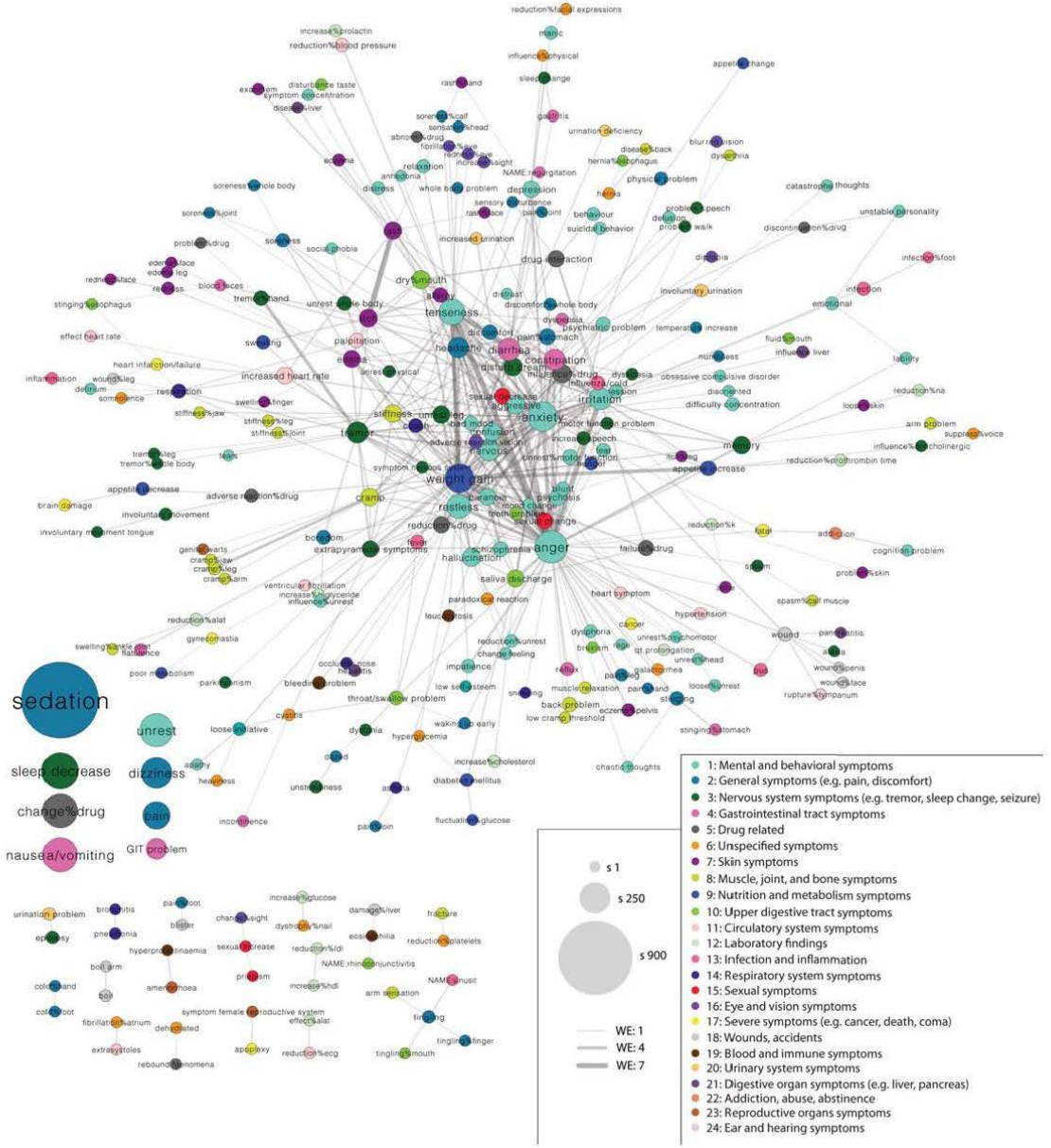
(ADRs text mined by temporal analysis of EHRs)

- N01 Anesthetics
- N02 Analgesics
- N03 Antiepileptics
- N04 Anti-parkinson drugs
- N05 Psycholeptics
- N06 Psychoanaleptics
- N07 Other nervous system drugs
- All other ATC codes

13 years of drug use at Mental centre Sct. Hans ATC code coloring. Edges show Drug ADR profile similarity, darker edge indicates stronger similarity. Network contains 500 strongest edges (Jaccard index). "N03 Antiepileptics" are scattered, not showing any clustering. This is expected as antiepileptics is a very diverse drug classein terms of ADRs. Laxatives and antibacterials for systemic use group since both cause diarrhea and stomach ache and other gastrointestinal problems

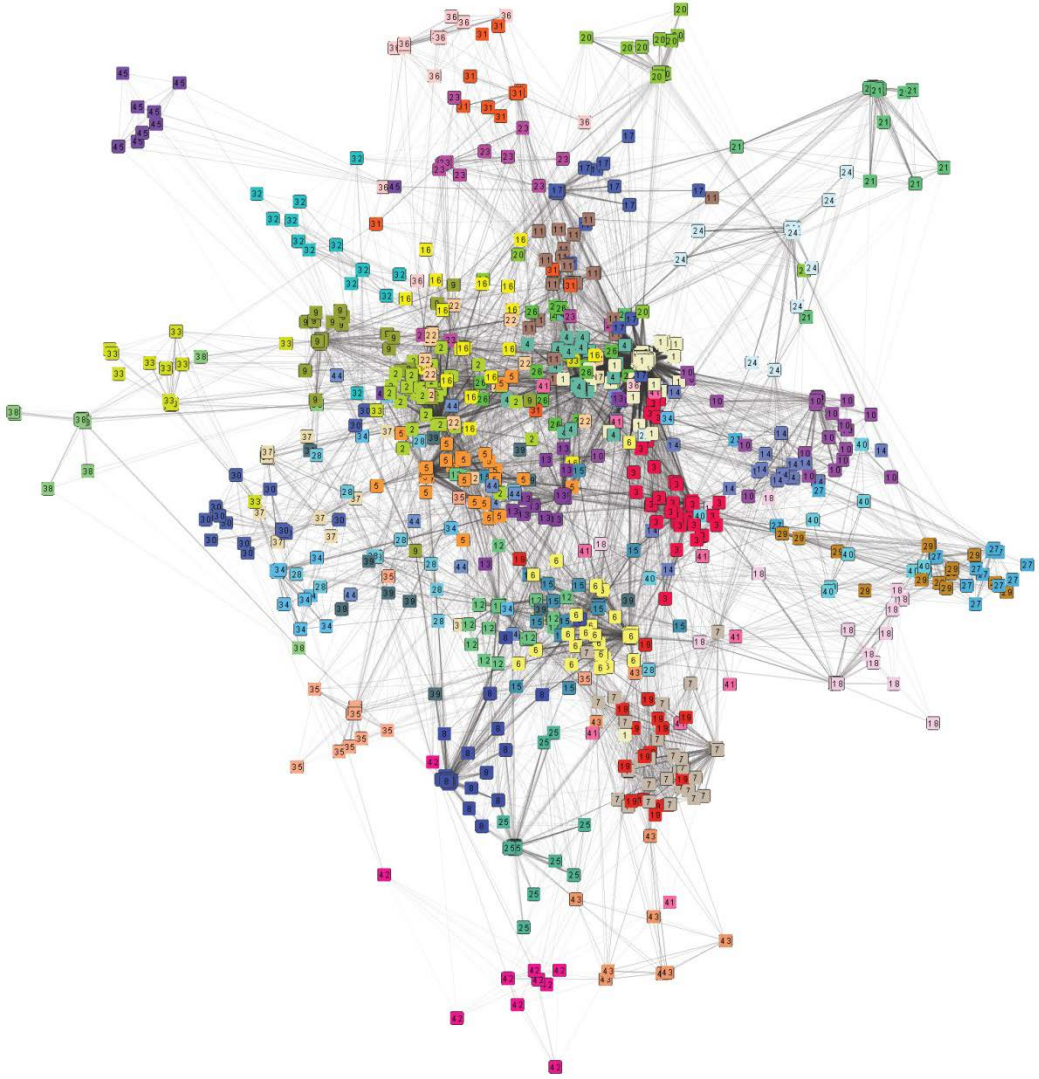


Co-occuring ADRs in epilesia patients



Roitmann et al.
Frontiers in Physiology
2014

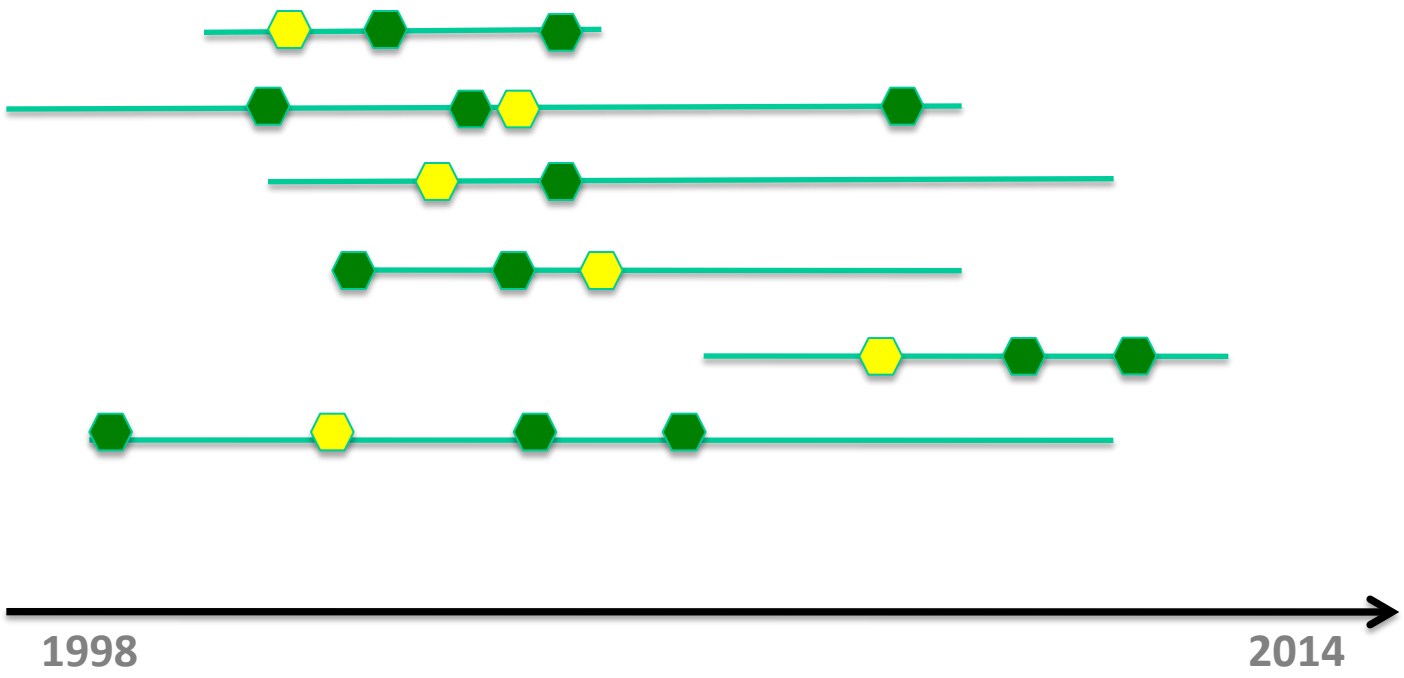
Epilesia patients clustered from their ADR vectors



Node color denotes cluster membership as determined by hierarchical clustering with a cutoff at 0.6.

Shown are only clusters with 10 or more patients and associations of a cosine dissimilarity value of less than 0.6.

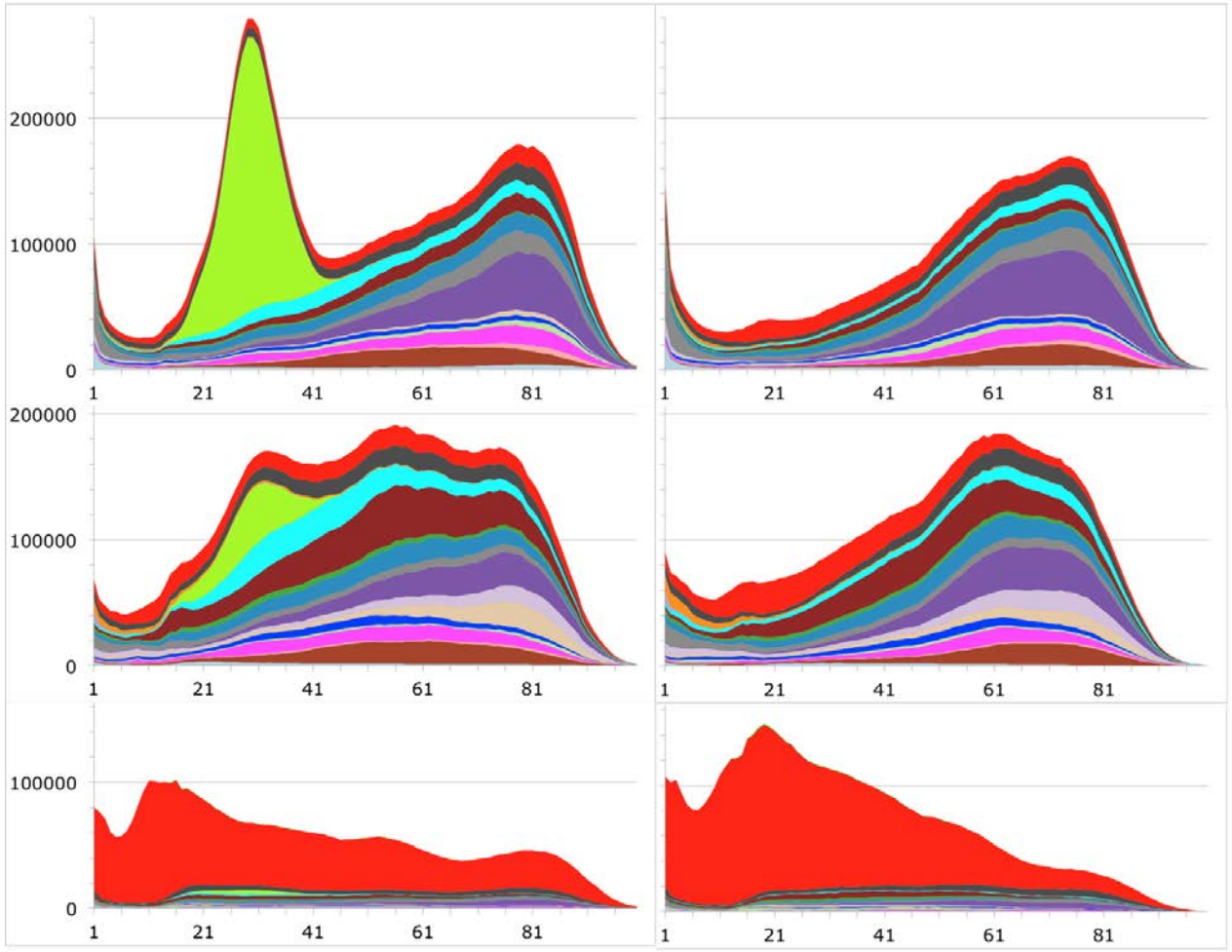
Comorbidity trajectories in individual patients



Typical development
E.g. type 2 diabetes > problems with foot > amputation

Yellow dot: Debut of a given ICD10 code, green debut of other diseases
Danish Discharge Registry, 6.2 million patients

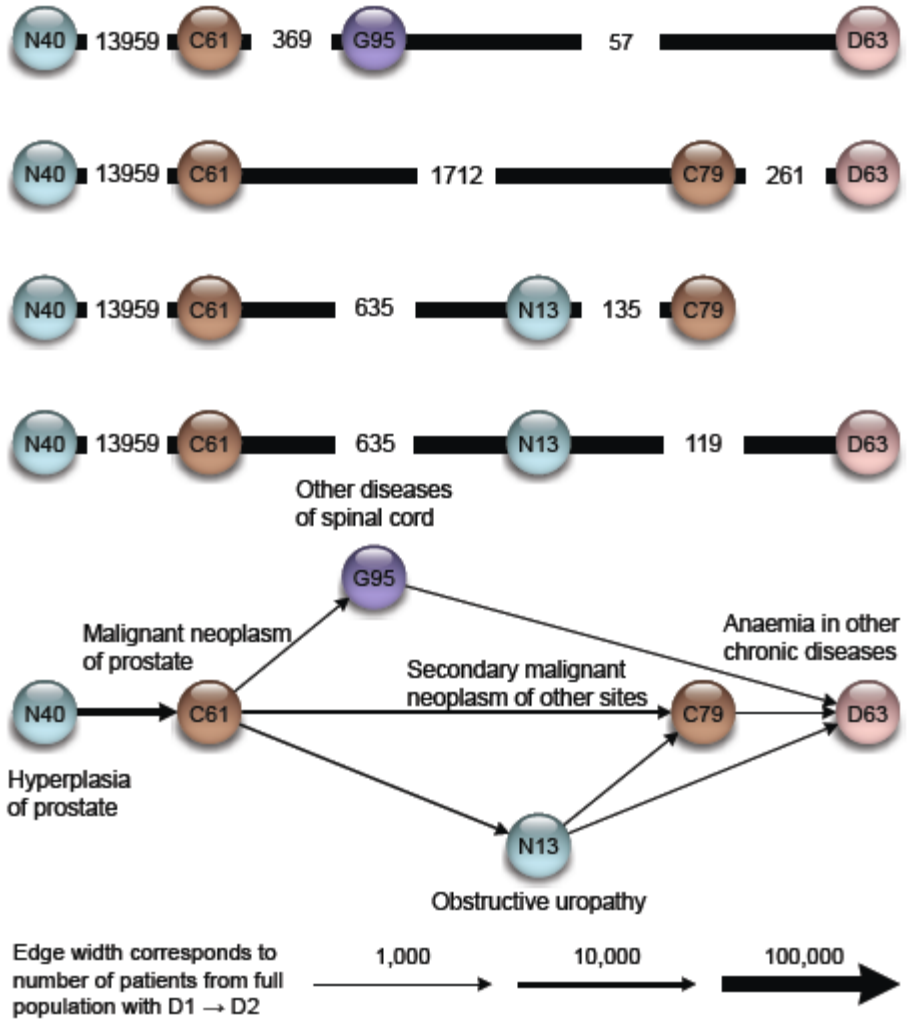
National Patient Registry (6.2M Danes) ICD10 diagnoses as a function of age



ICD 10 chapter coloring

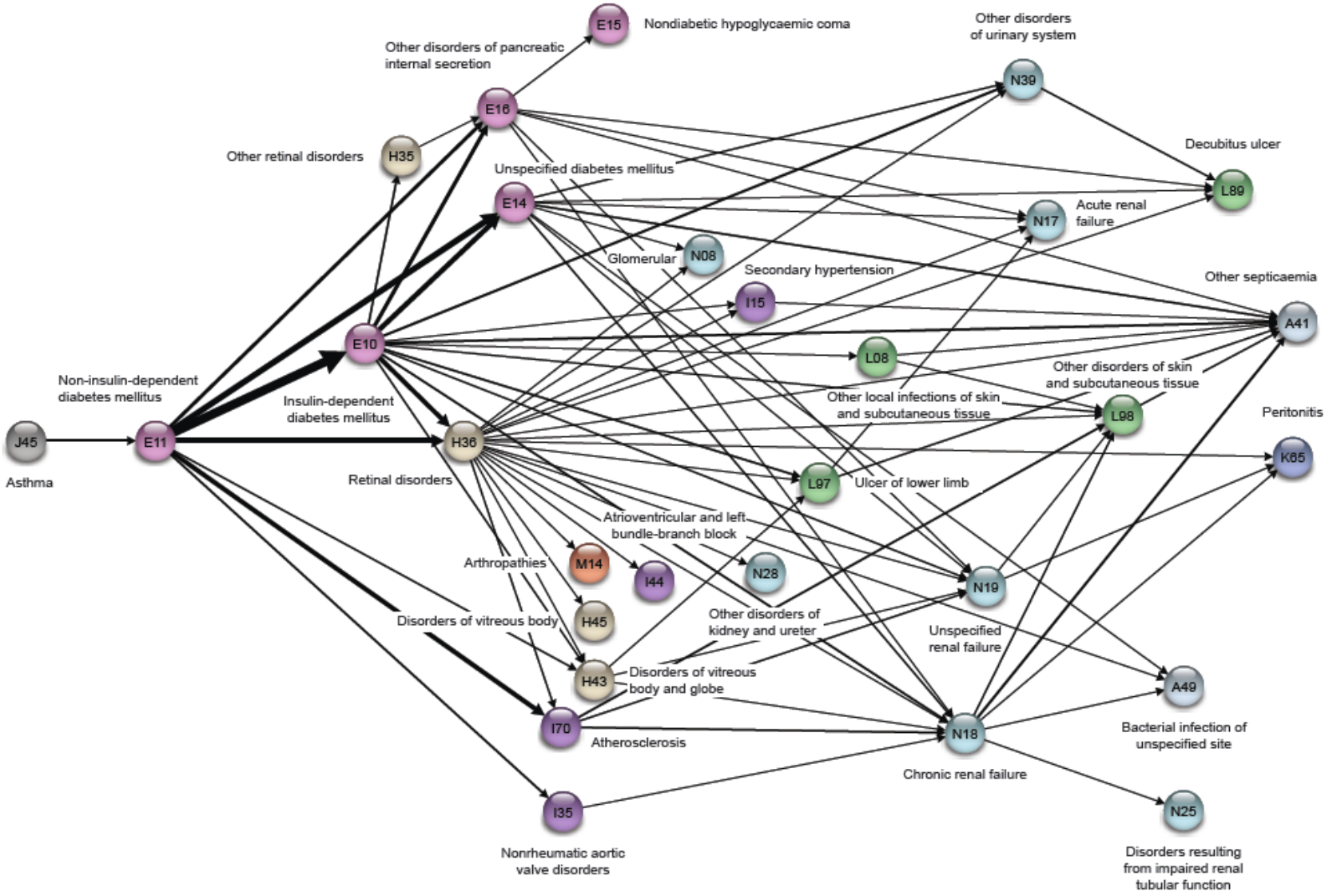
1: Certain infectious and parasitic diseases
2: Neoplasms
3: Diseases of the blood and blood-forming organs and certain disorders involving the immune mechanism
4: Endocrine, nutritional and metabolic diseases
5: Mental and behavioural disorders
6: Diseases of the nervous system
7: Diseases of the eye and adnexa
8: Diseases of the ear and mastoid process
9: Diseases of the circulatory system
10: Diseases of the respiratory system
11: Diseases of the digestive system
12: Diseases of the skin and subcutaneous tissue
13: Diseases of the musculoskeletal system and connective tissue
14: Diseases of the genitourinary system
15: Pregnancy, childbirth and the puerperium
16: Certain conditions originating in the perinatal period
17: Congenital malformations, deformations and chromosomal abnormalities
18: Symptoms, signs and abnormal clinical and aboratory findings, not elsewhere classified
19: Injury, poisoning and certain other consequences of external causes
20: External causes of morbidity and mortality

Disease trajectories and trajectory-cluster for prostate cancer



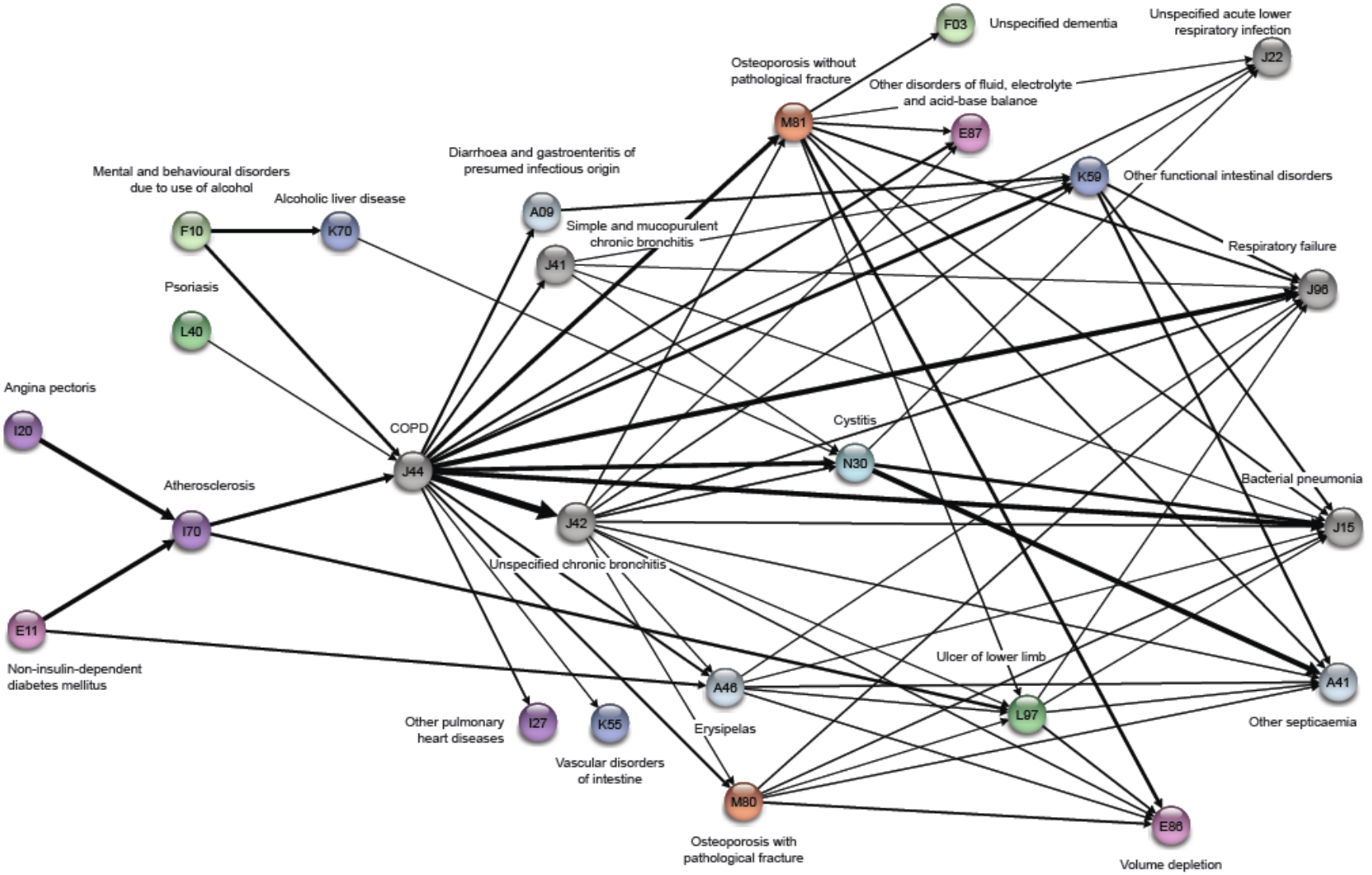
AB Jensen et al., Nature Comm., 2014

Diabetes trajectory network

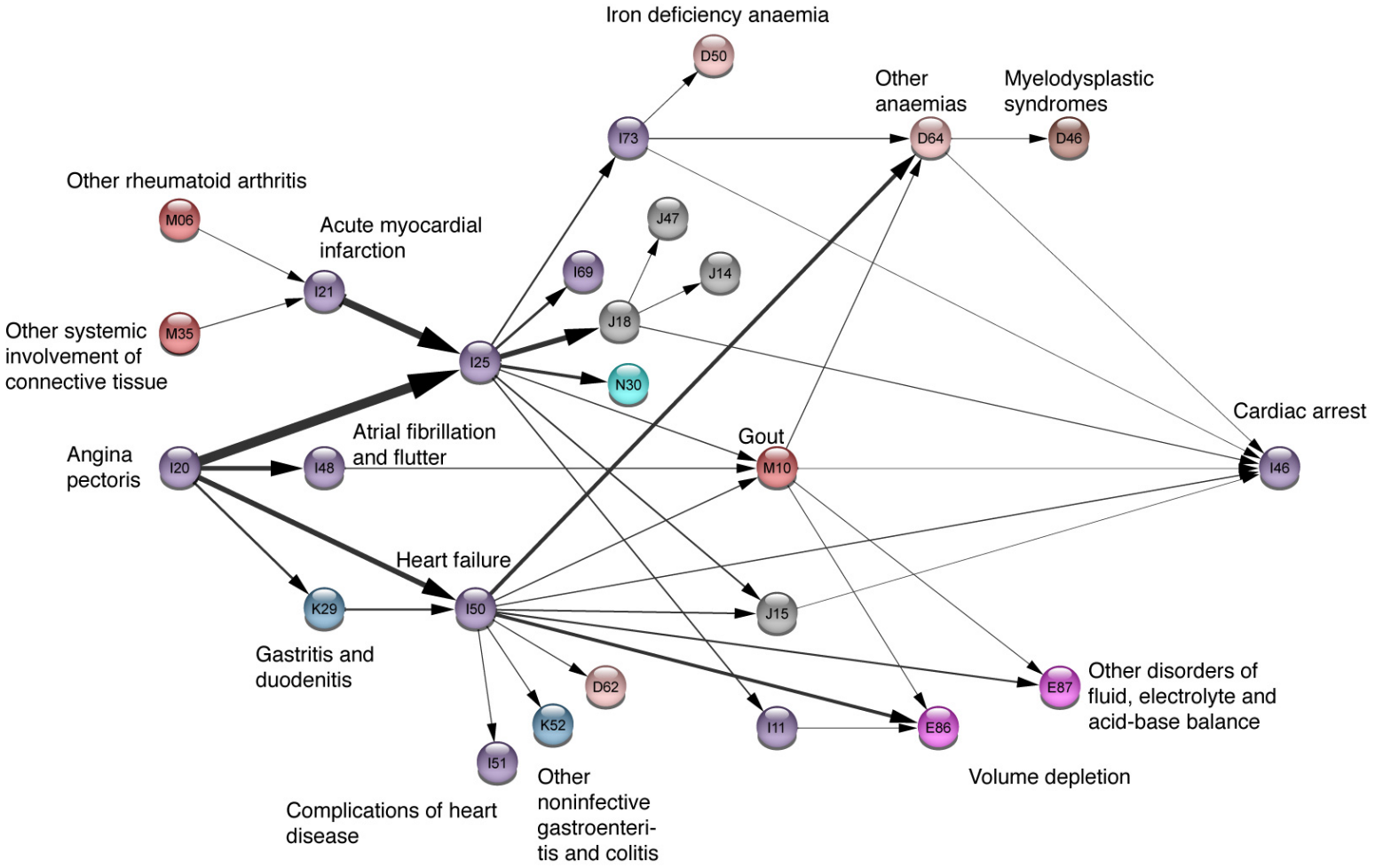


COPD trajectory cluster

with five preceding diagnoses leading to COPD and some of the possible outcomes



Cardiovascular trajectory network



Musings

Highly accessed

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

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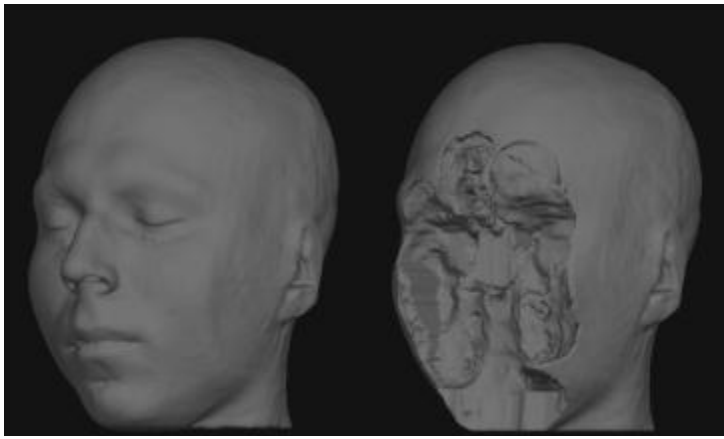
De-identification

In the Nordic countries and in some other countries we can minimize the effort on rediscovering what we already know



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SEVENTH FRAMEWORK PROGRAMME

Danish Agency for Science Technology and Innovation
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TRANSLATIONAL GENETICS

Mining electronic health records: towards better research applications and clinical care

Peter B. Jensen[1], Lars J. Jensen[1] and Søren Brunak[1,2]

Abstract | Clinical data describing the phenotypes and treatment of patients represents an underused data source that has much greater research potential than is currently realized. Mining of electronic health records (EHRs) has the potential for establishing new patient-stratification principles and for revealing unknown disease correlations. Integrating EHR data with genetic data will also give a finer understanding of genotype–phenotype relationships. However, a broad range of ethical, legal and technical reasons currently hinder the systematic deposition of these data in EHRs and their mining. Here, we consider the potential for furthering medical research and clinical care using EHR data and the challenges that must be overcome before this is a reality.

Clinical decision support (CDS). Software systems providing support for decision making to physicians through the application of health knowledge and logical rules to patient data.

Biobanks Central repositories of biological material that are mainly used for research. They facilitate the re-use of collected samples in different research projects.

Information technology has transformed the way health care is carried out and documented. Presently, the practice of health care generates, exchanges and stores huge amounts of patient-specific information. In addition to the traditional clinical narrative, databases in modern health centres automatically capture structured data relating to all aspects of care, including diagnosis, medication, laboratory test results and radiological imaging data.

This transformation holds great promise for the individual patient as richer information, coupled with clinical decision support (CDS) systems, becomes readily available at the bedside to support informed decision-making and to improve patient safety[1,2]. It is especially interesting when traditional health-care-sector data is linked with biobanks and genetic data[4].

Despite the great potential, researchers who wish to analyse large amounts of patient data are still faced with technical challenges of integrating scattered, heterogeneous data, in addition to ethical and legal obstacles that limit access to the data[5,6]. It is hoped that large-scale adoption of health information technology (HIT) infrastructure in the form of electronic health records (EHRs) and agreed standards for interoperability and schemes for privacy and consent, will improve this situation (TABLE 1). With incentives for improved public health and the expected health budget savings[7,8], these matters