



INFOBIOMED

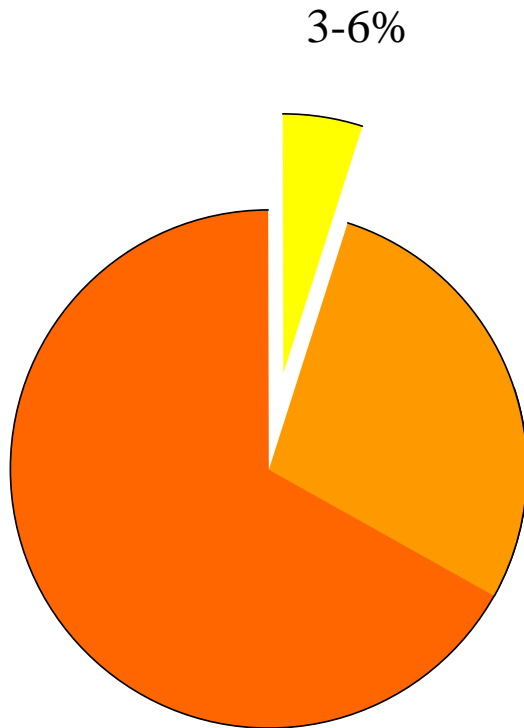
Genomics and colon cancer

Electronic data exchange

Inge Bernstein

The Danish HNPCC-register, Hvidovre Hospital, Denmark

Hereditary Colon cancer



HNPCC : 3-6% of CRC

Inherited risk CRC: up to 35%

HNPCC identification & screening

Reduced incidence CRC $> 60\%$

Earlier detection CRC

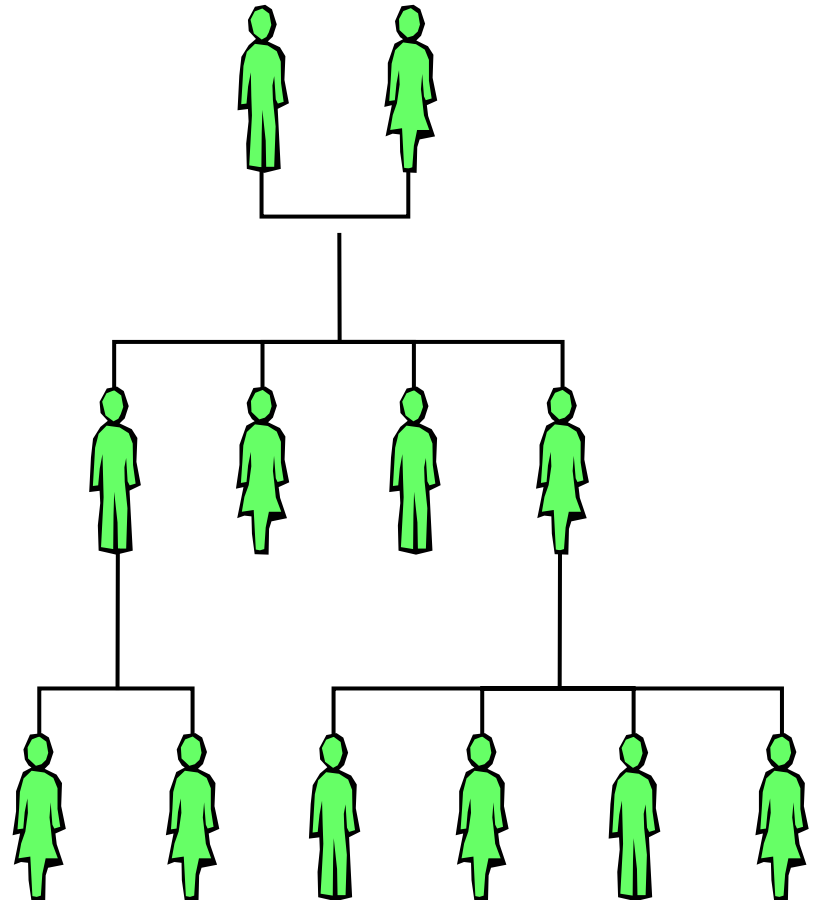
Better survival CRC

Cost-effective

Lifetime risk - CRC

Sporadic

4%



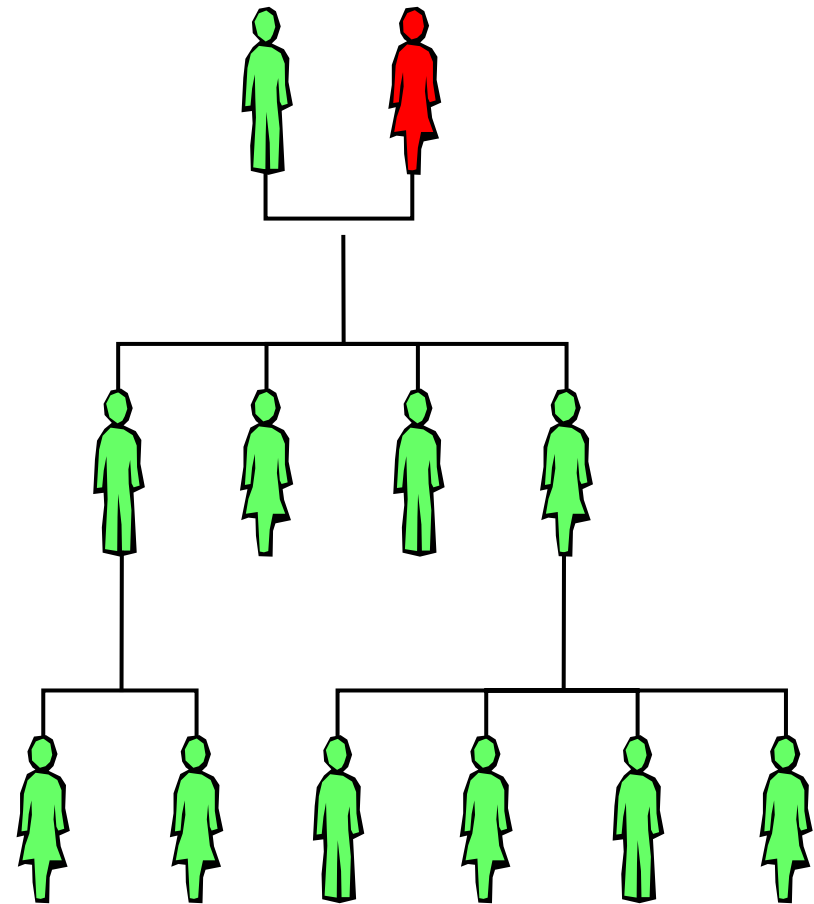
Lifetime risk - CRC

Sporadic

4%

One relative

6%



Lifetime risk - CRC

Sporadic

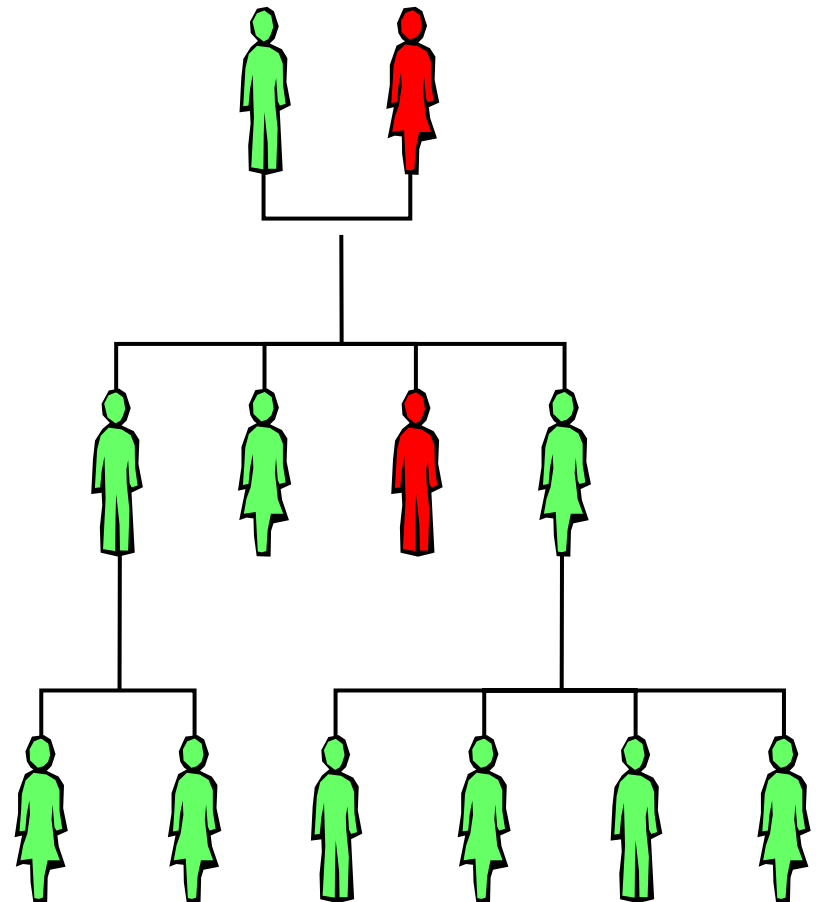
4%

One relative

6%

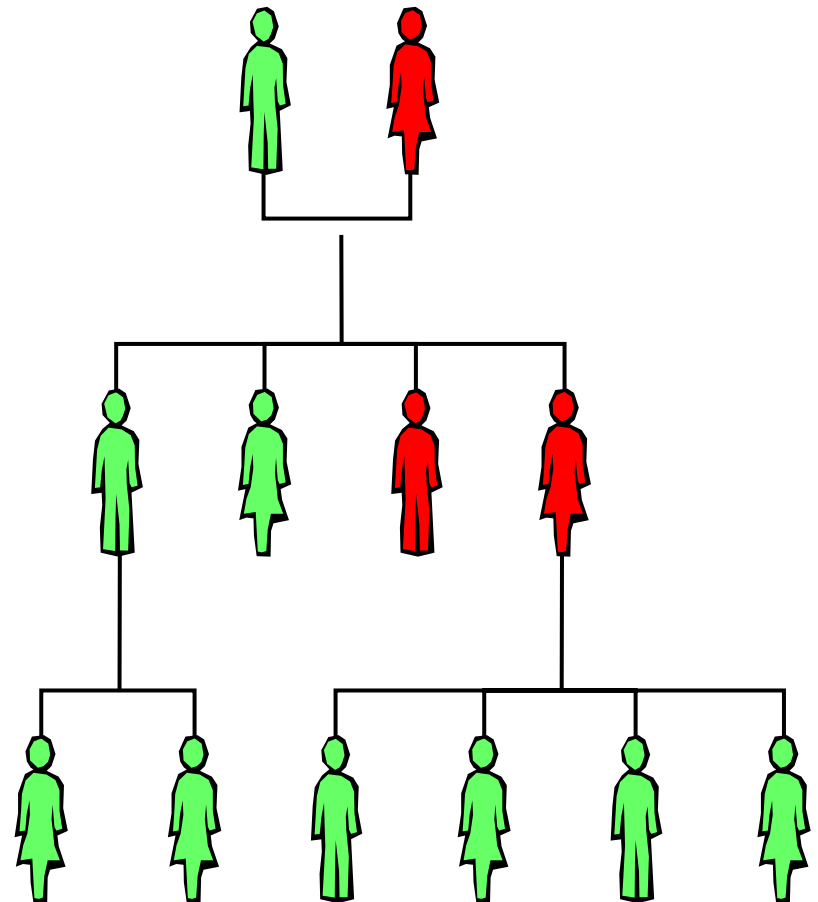
Two relatives

17%



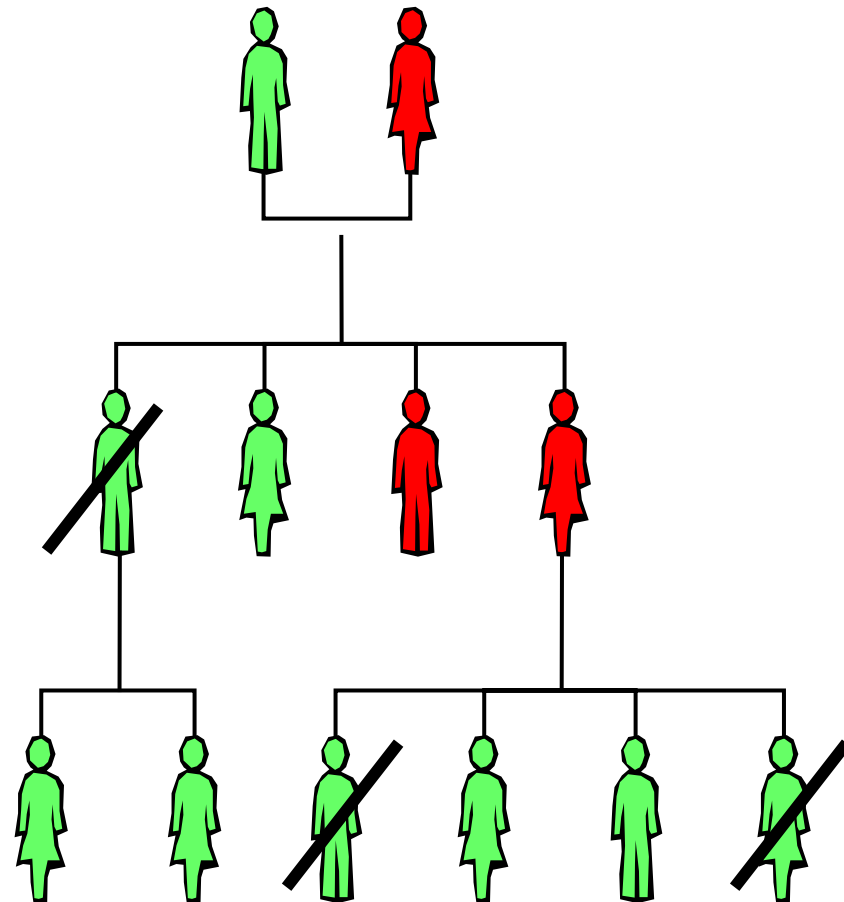
Lifetime risk - CRC

Sporadic	4%
One relative	6%
Two relatives	17%
At risk HNPCC	40-45%



Lifetime risk - CRC

Sporadic	4%
One relative	6%
Two relatives	17%
At risk HNPCC	40-45%
Gene carrier HNPCC	80-90%



Phenotype – Genotype

Family level

Pedigree

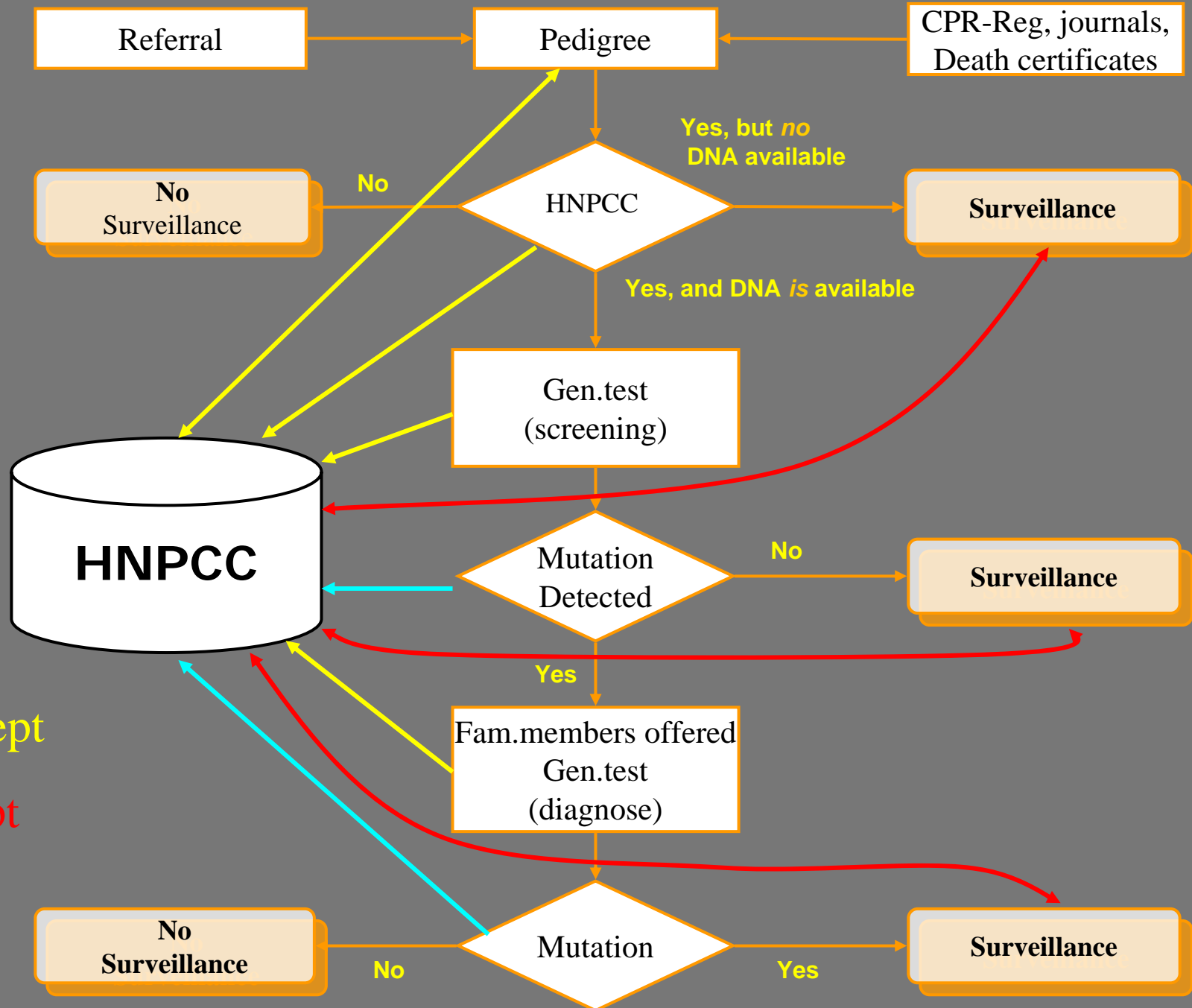
Individual level

Cancers – prelesions

Mutations – SNP

Risk estimation – surveillance programs

Data flow



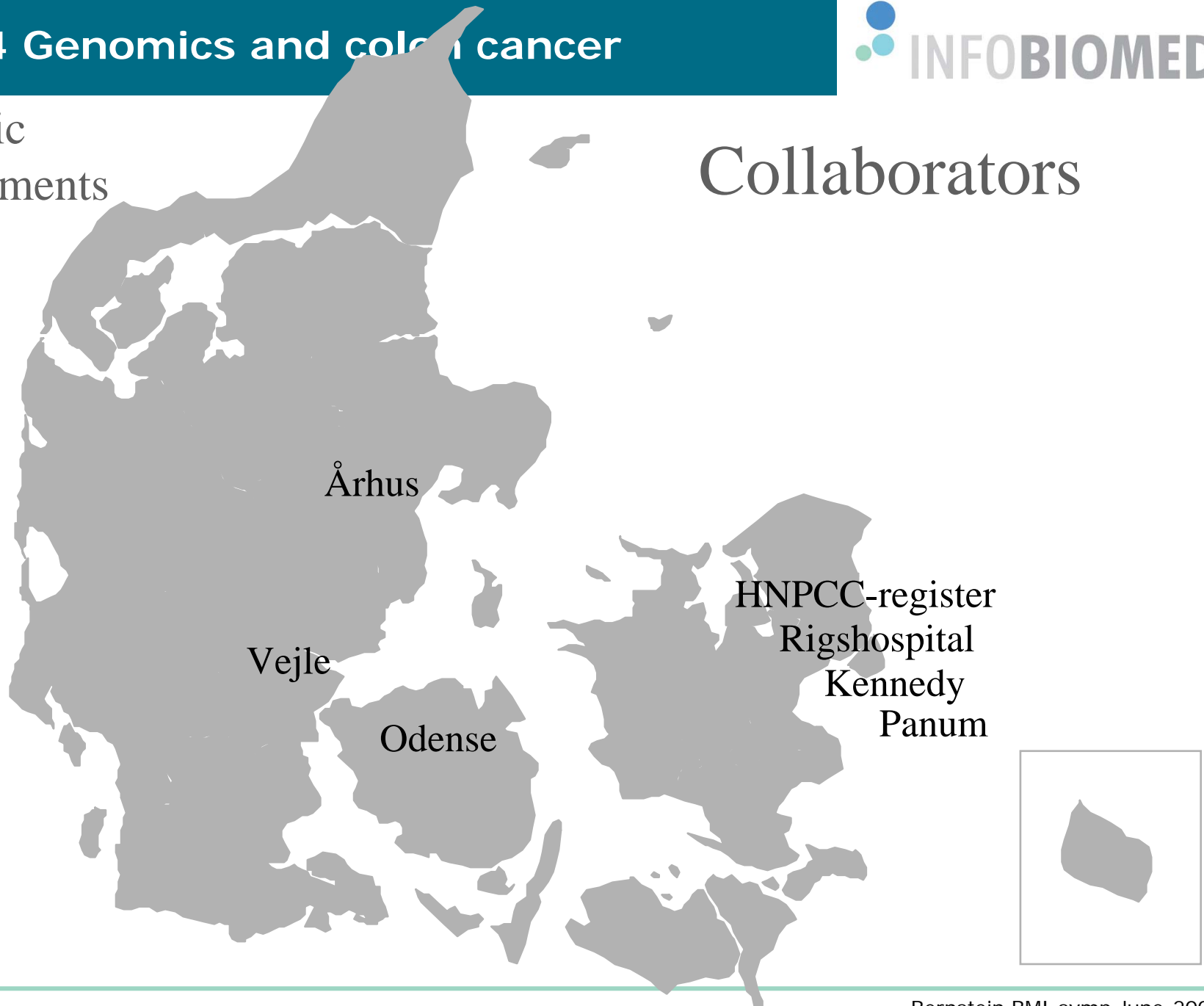
Gen. dept

Sur.dept

Lab.

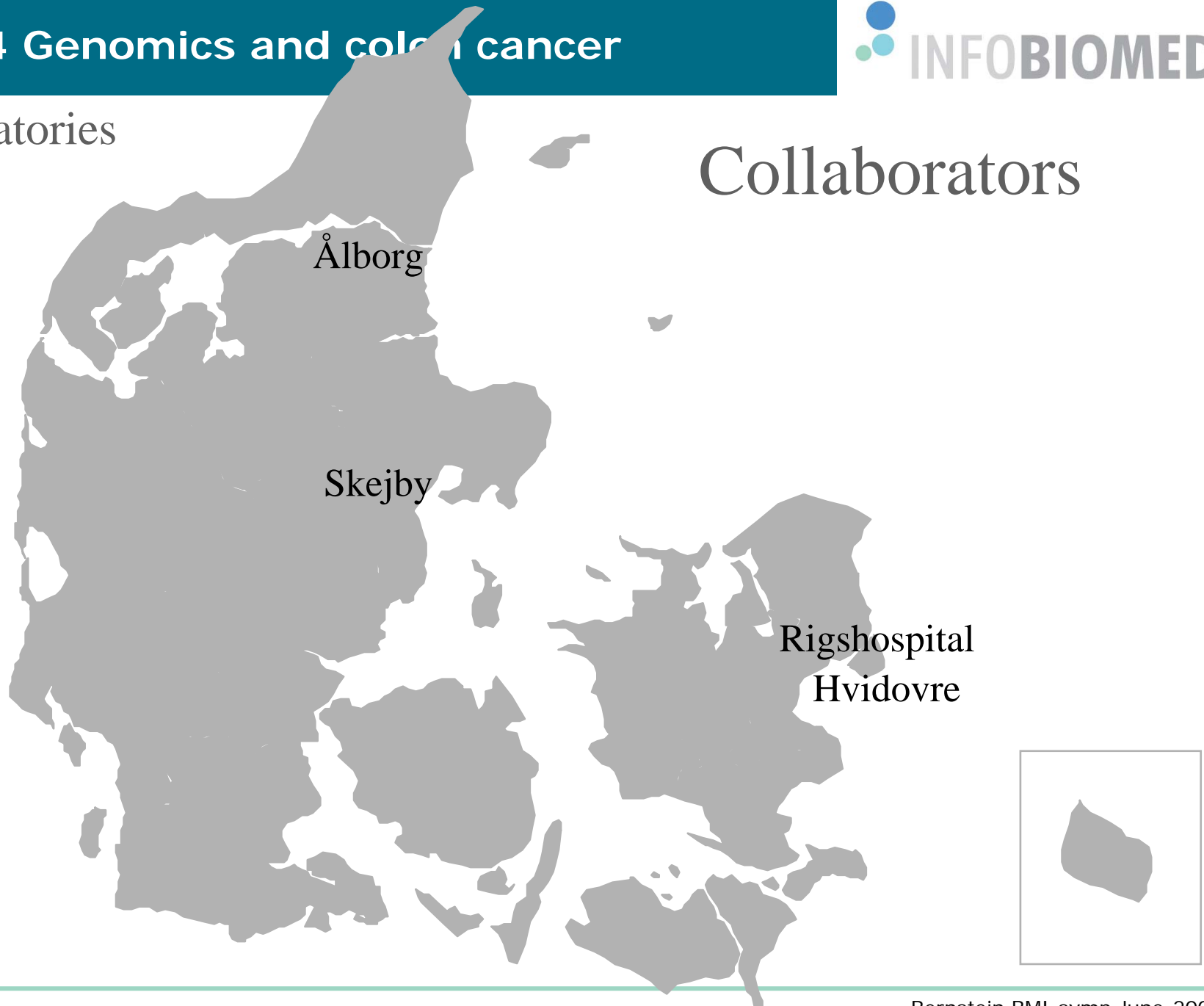
Genetic
departments

Collaborators



Laboratories

Collaborators



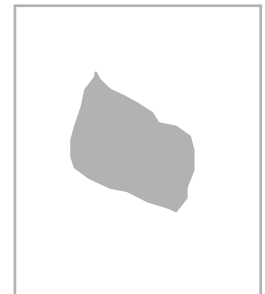
Surgical
departments

Collaborators



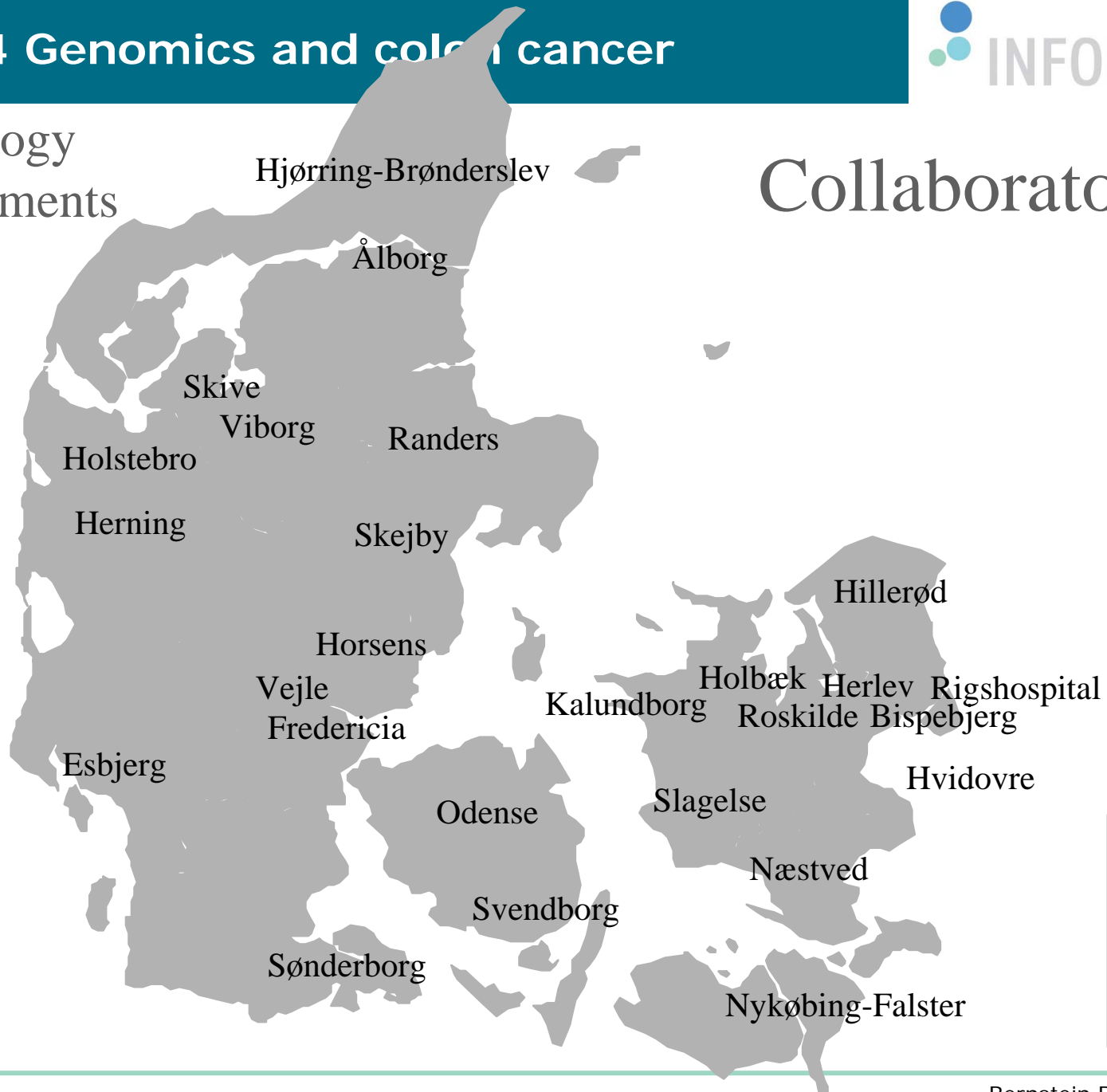
Gynaecologic
departments

Collaborators



Pathology
departments

Collaborators



HNPCC-register

*Today:
Paper based*



*6 genetic dept.
4 laboratories
40 surg. dept.
> 30 gyn. dept.
25 path. dept*

Future Aims:

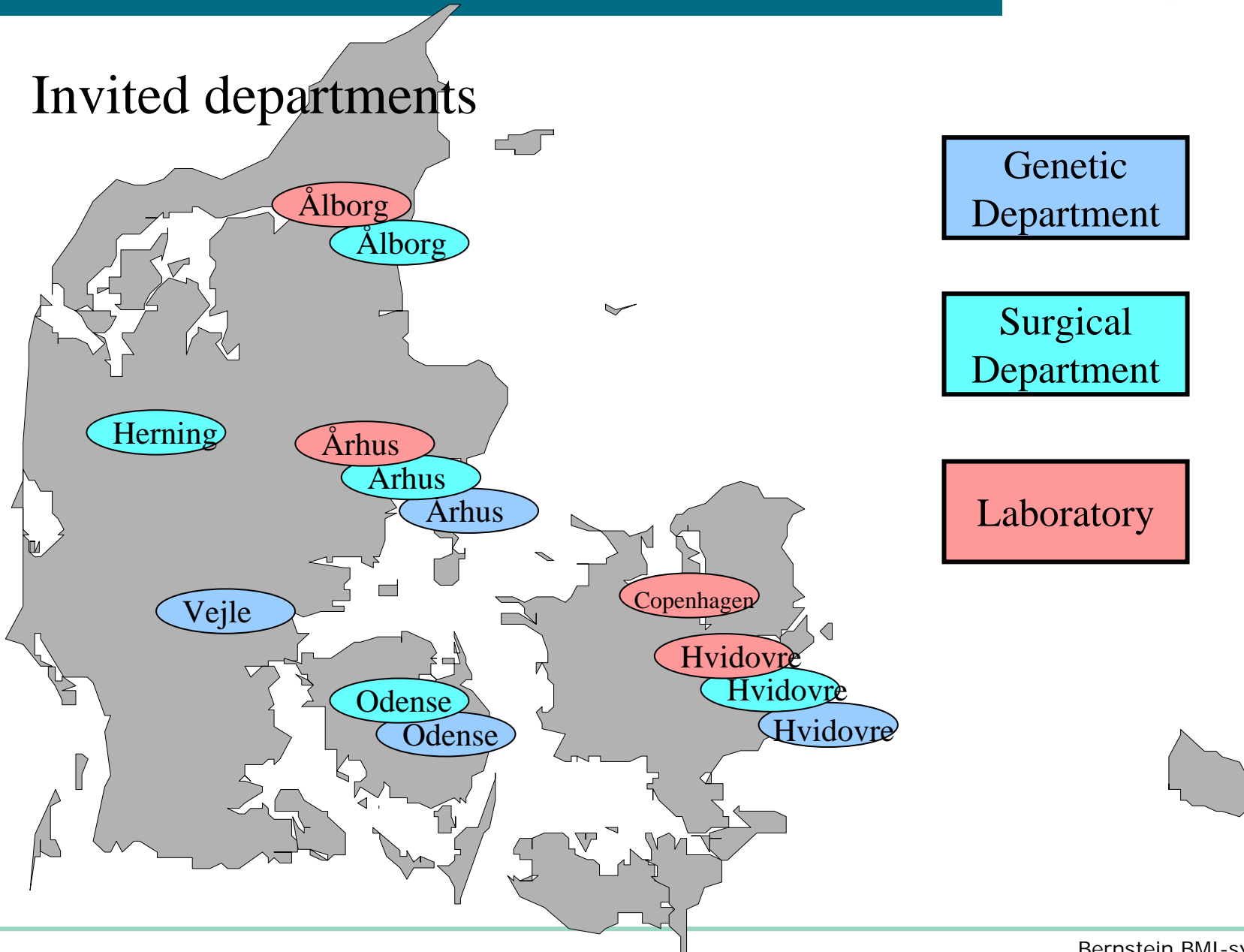
Interoperability

Communication

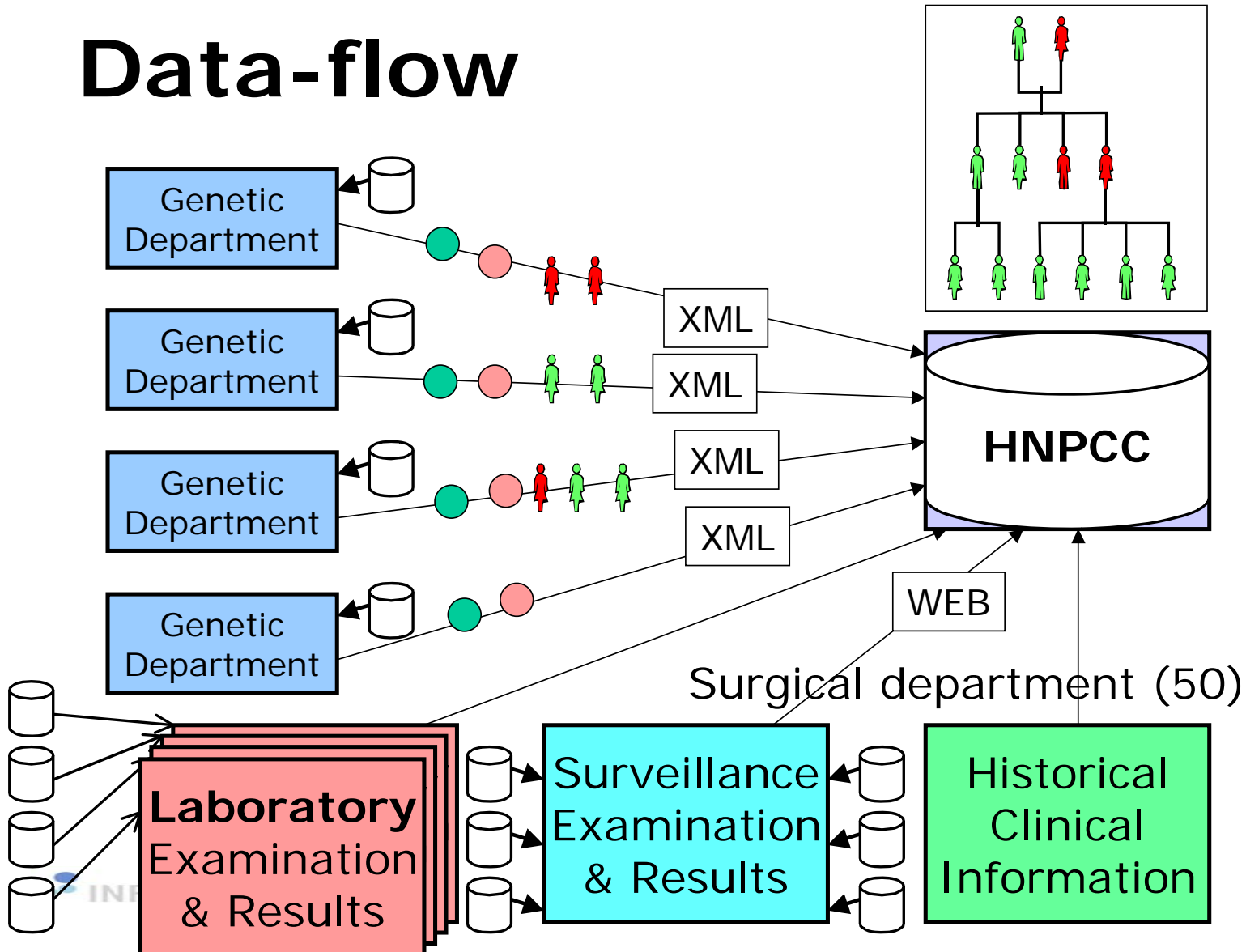
Further IT-support

Web-reporting of data

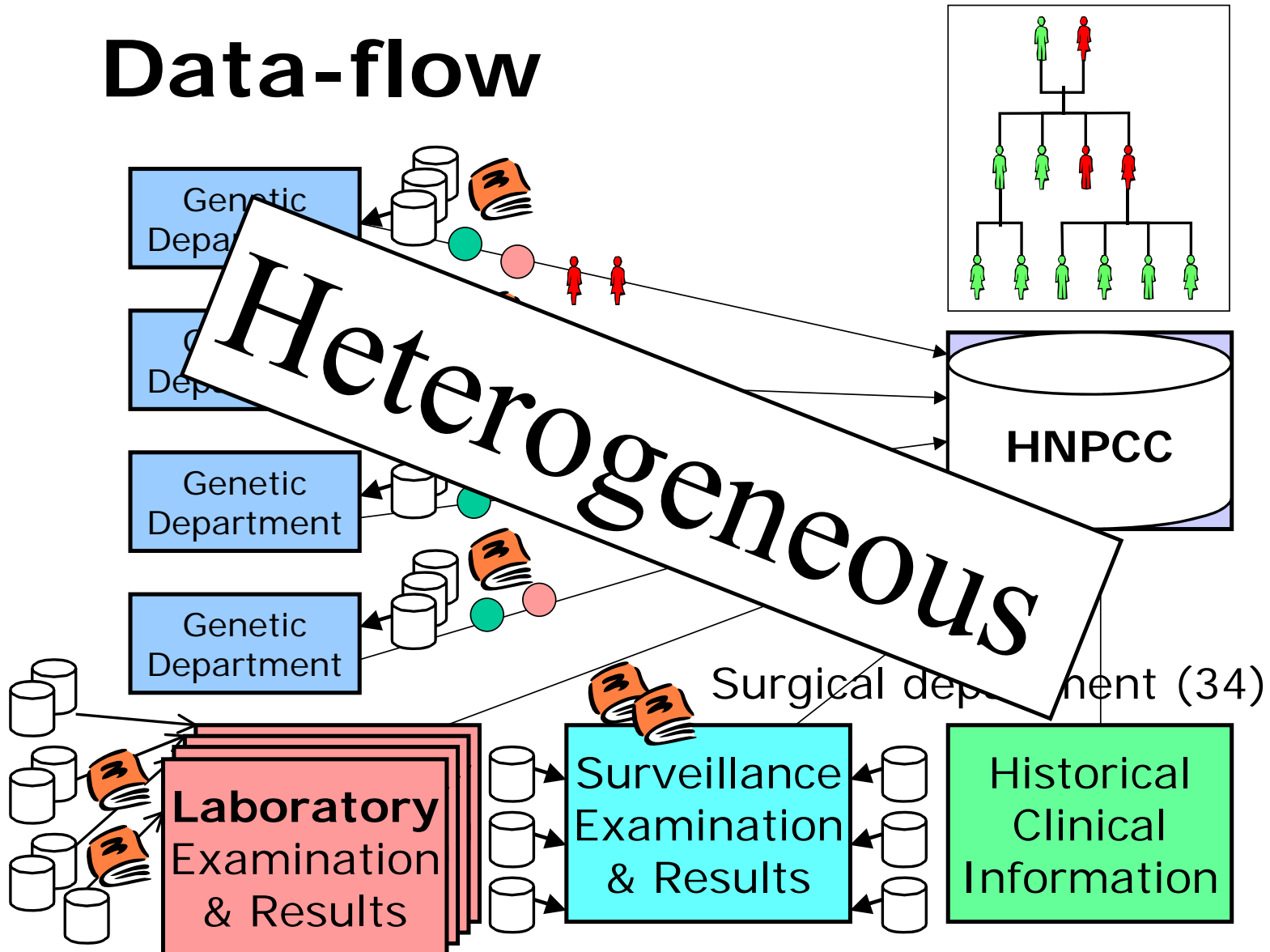
Invited departments



Data-flow



Data-flow

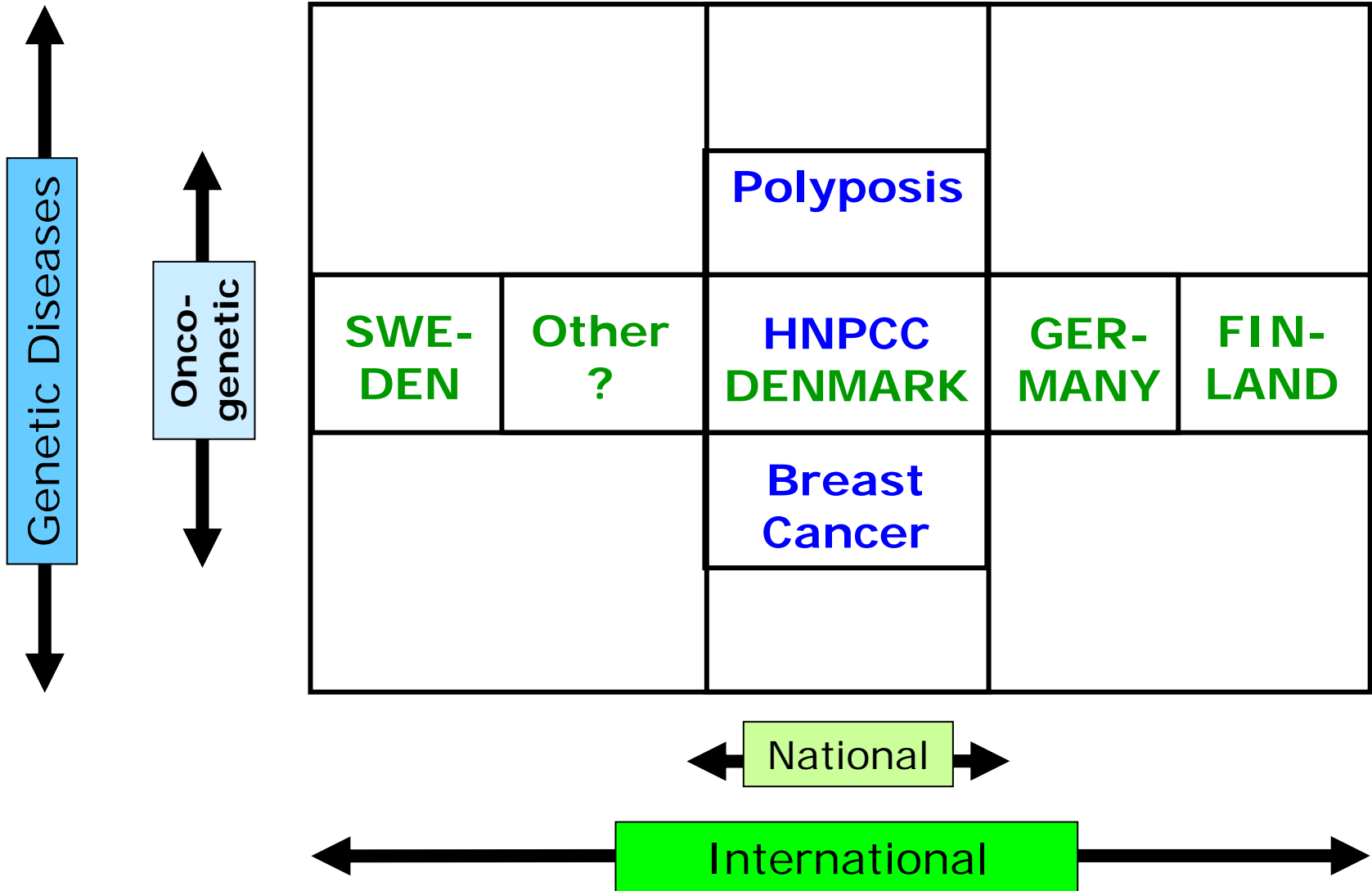


HNPPC - standards

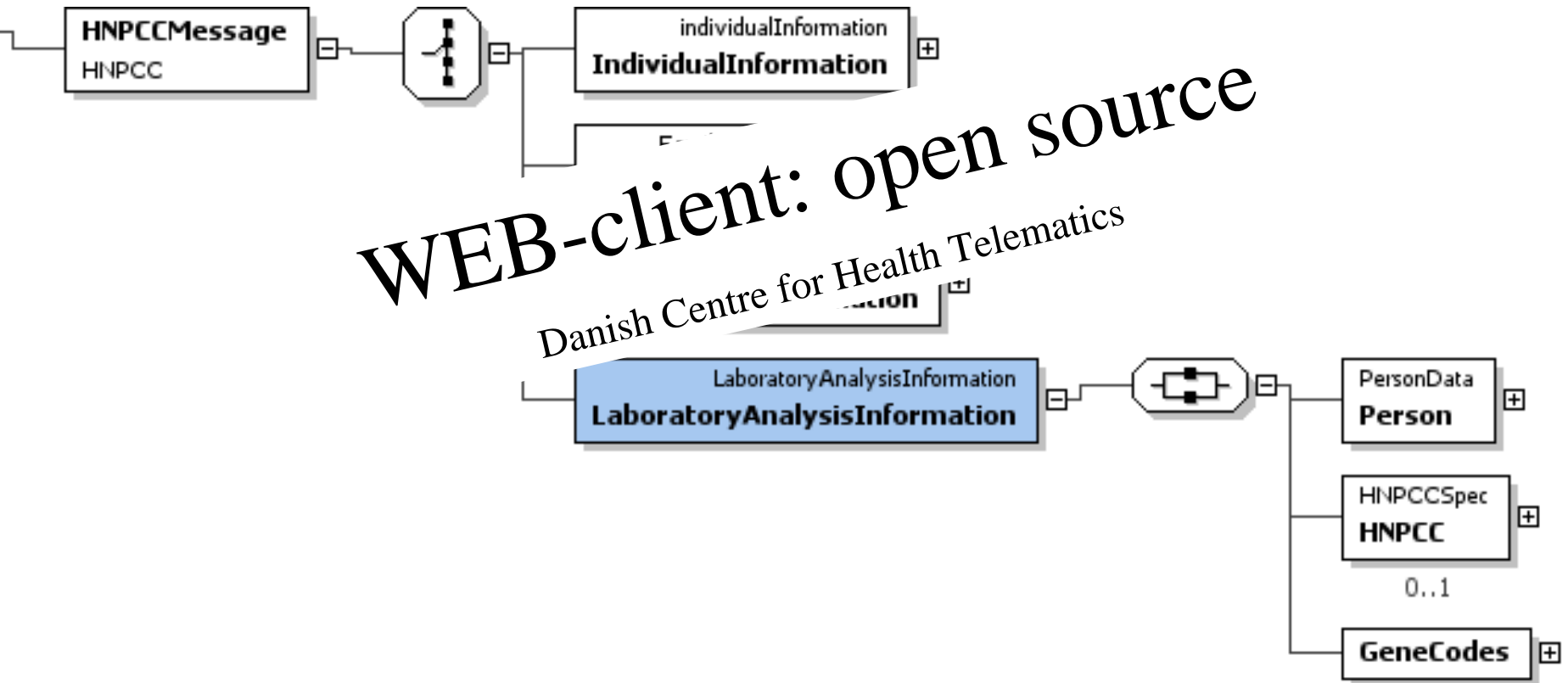
- Family data - Phenotype
- Individual data - Phenotype
- Mutations/SNPs - Genotype
- Surveillance - Phenotype



INFOBIOMED

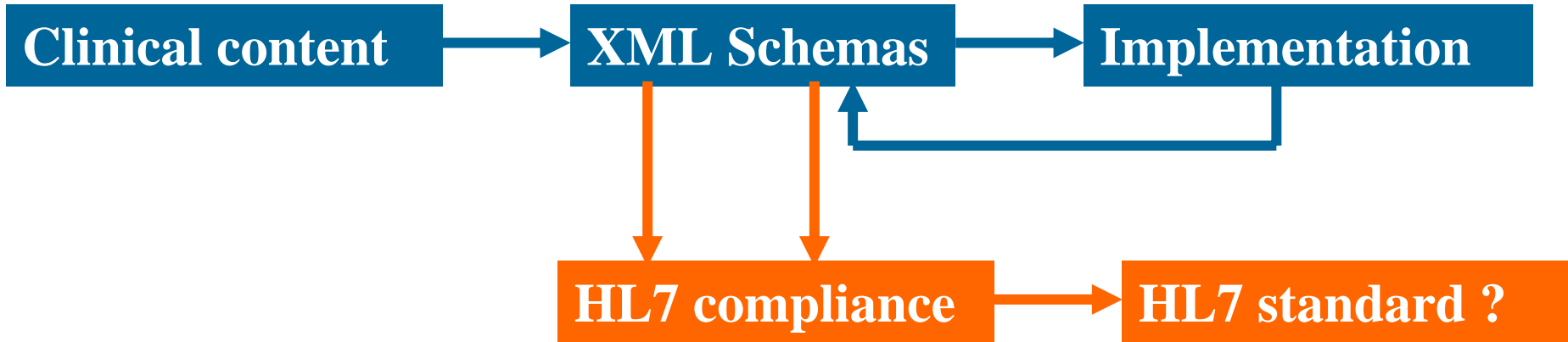


Design of XML documents



WEB-client: open source

Danish Centre for Health Telematics



WEB-client

Report Statistic About Log Out

Report type: Surveillance Patient name: Max Bergren
Cpr-nr: 100270-1345

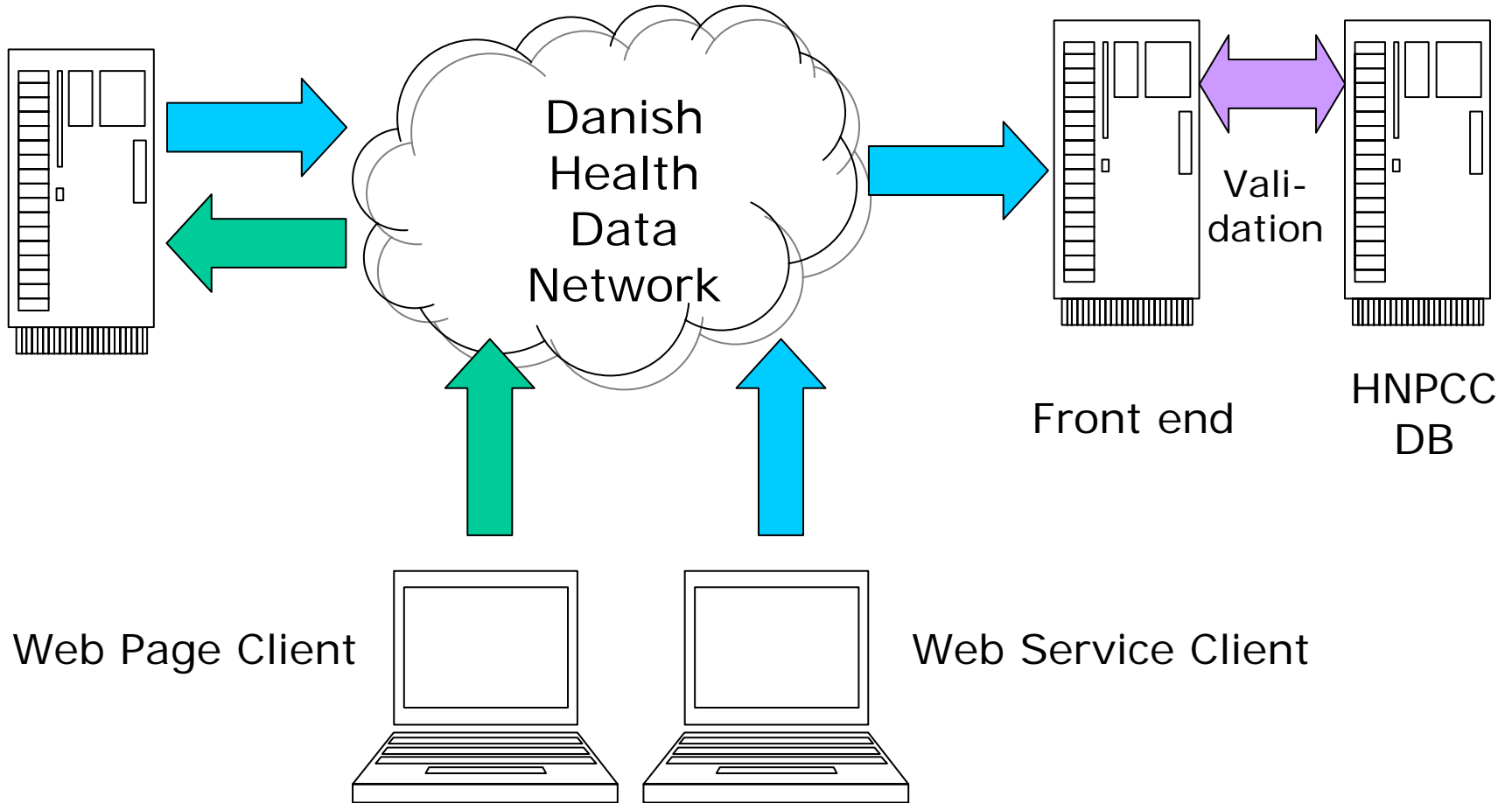
- Patient
- HNPCC
- Treatment
- Results
 - Result
 - Cancers
 - Cancer
 - Cancer
 - Cancer
 - Result
 - Result
- Send Report
- Cancel Report

Cancer	✕ Delete
ICD Code	<input type="text" value="1531"/>
Description	<input type="text" value="Transverse colon"/>

HNPCC-registret

Web Server


Web Service Provider



Laboratory / Surgery Dep. / Genetic Dep.

 HTML

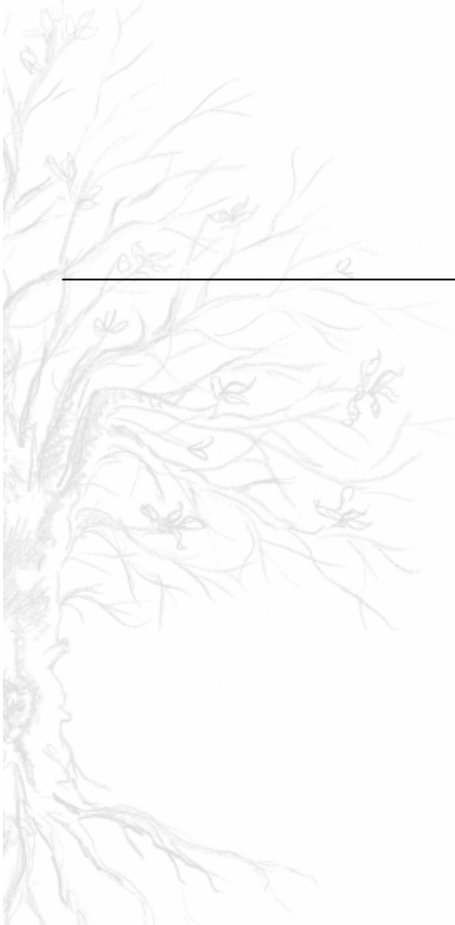
 Web Service

 Not part of InfoBioMed

Login

Login	
Username	<input type="text" value="kir07ber"/>
Password	<input type="password" value="....."/>
<input type="button" value="Login"/>	

The HNPCC-register

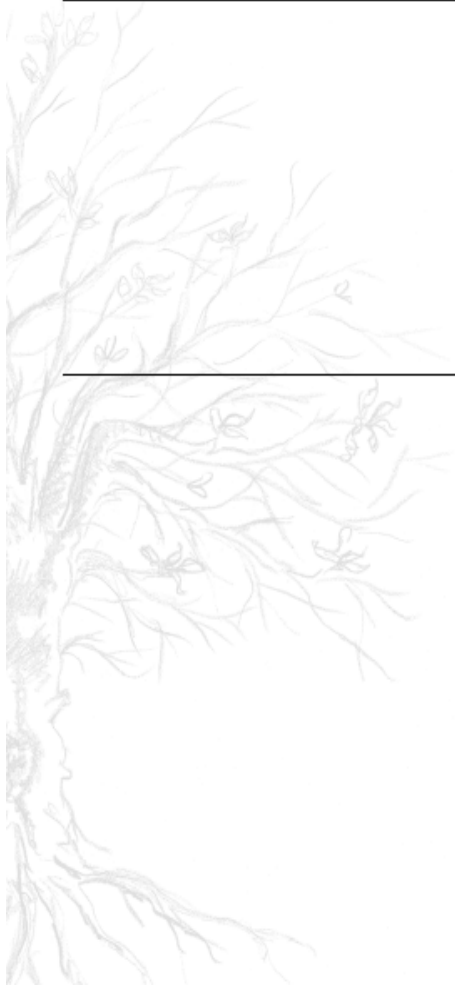


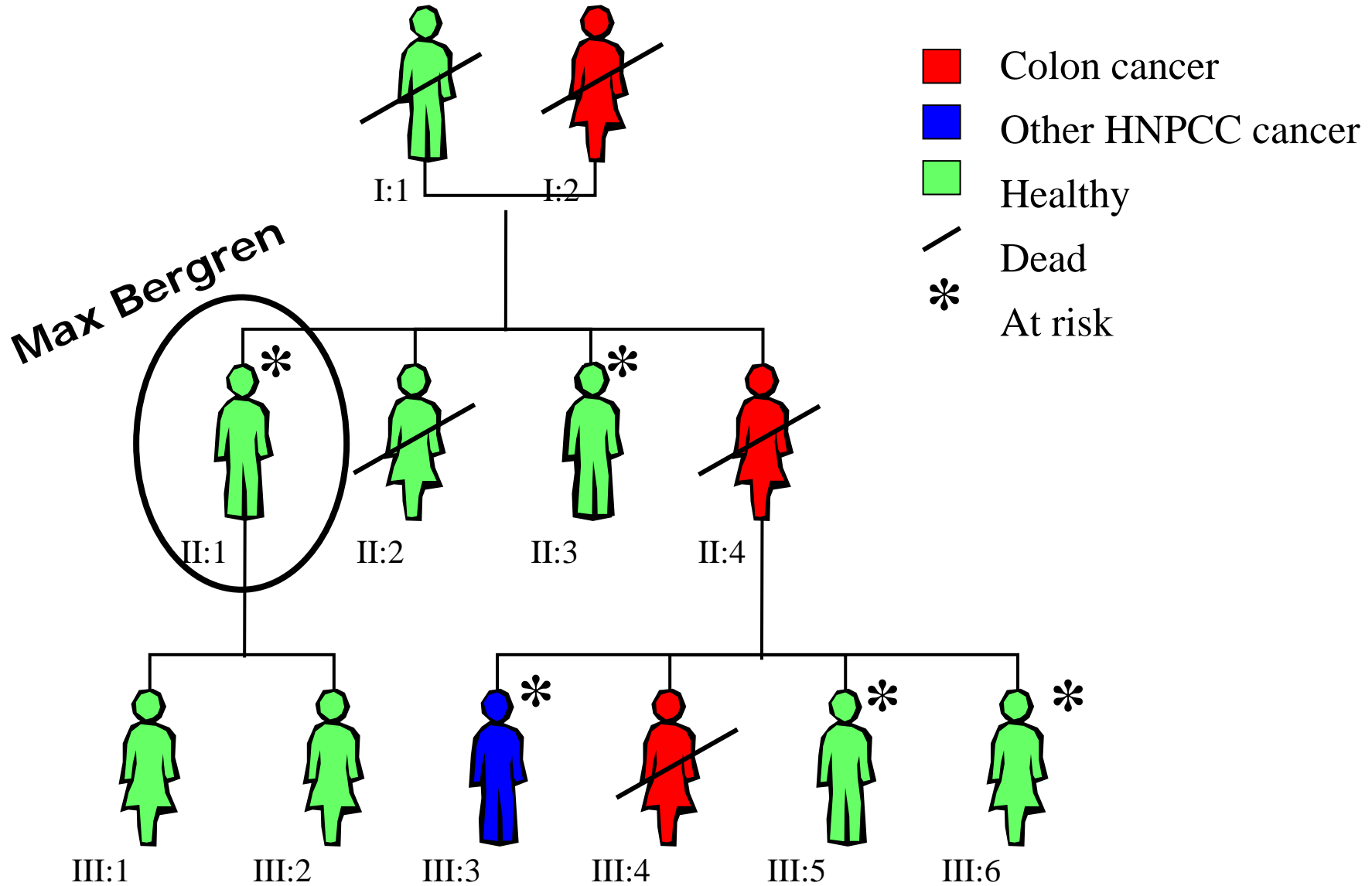
Report Statistics About Log Out

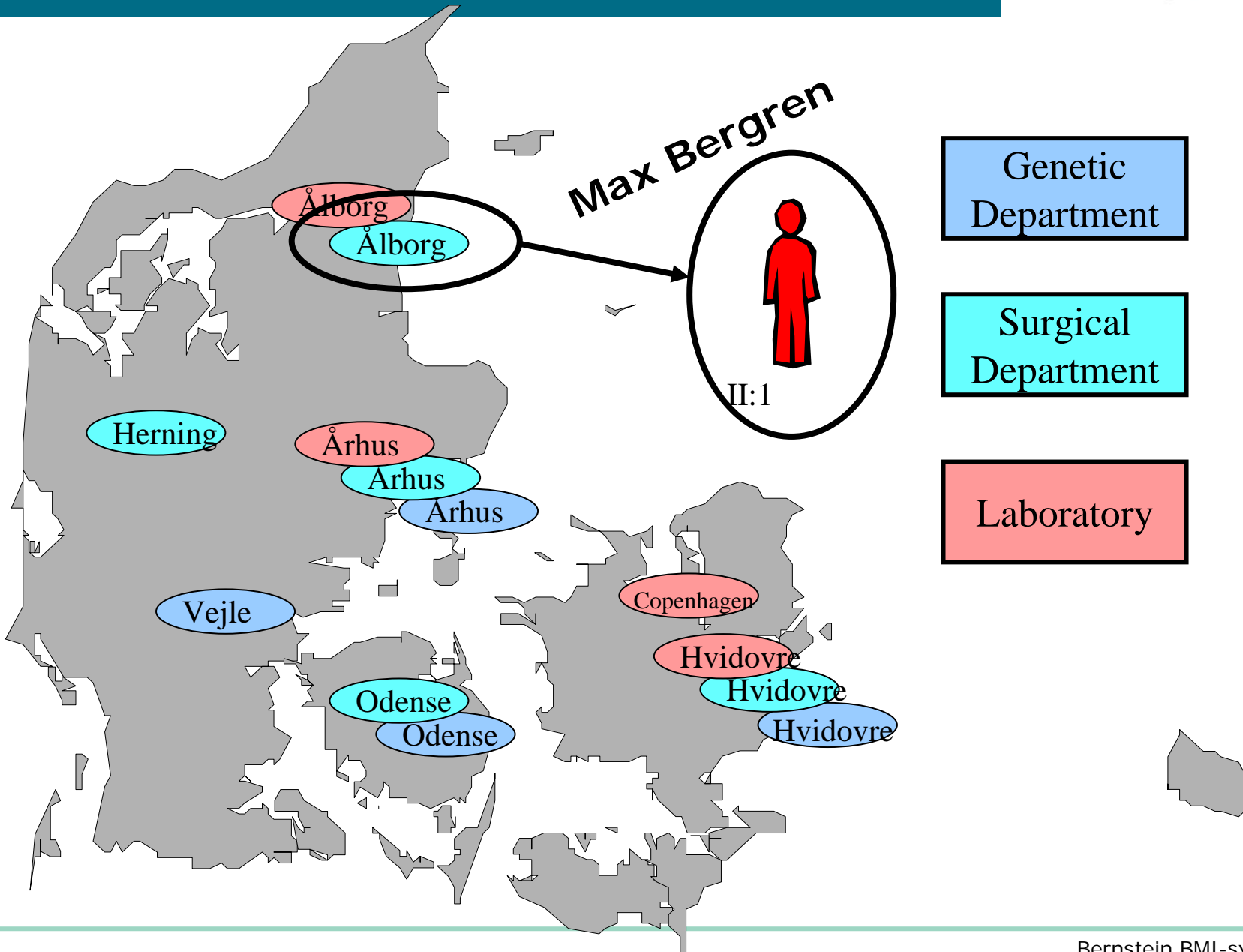
Select type of report to send

- Family
- Individual
- Laboratory Analysis
- Surveillance

The HNPCC-register







Login

Login Form	
Username	<input type="text" value="Bernstein"/>
Password	<input type="password" value="....."/>
<input type="button" value="Login"/>	

HNPCC-registret


Report Statistic About Log Out

Report type: **Surveillance**

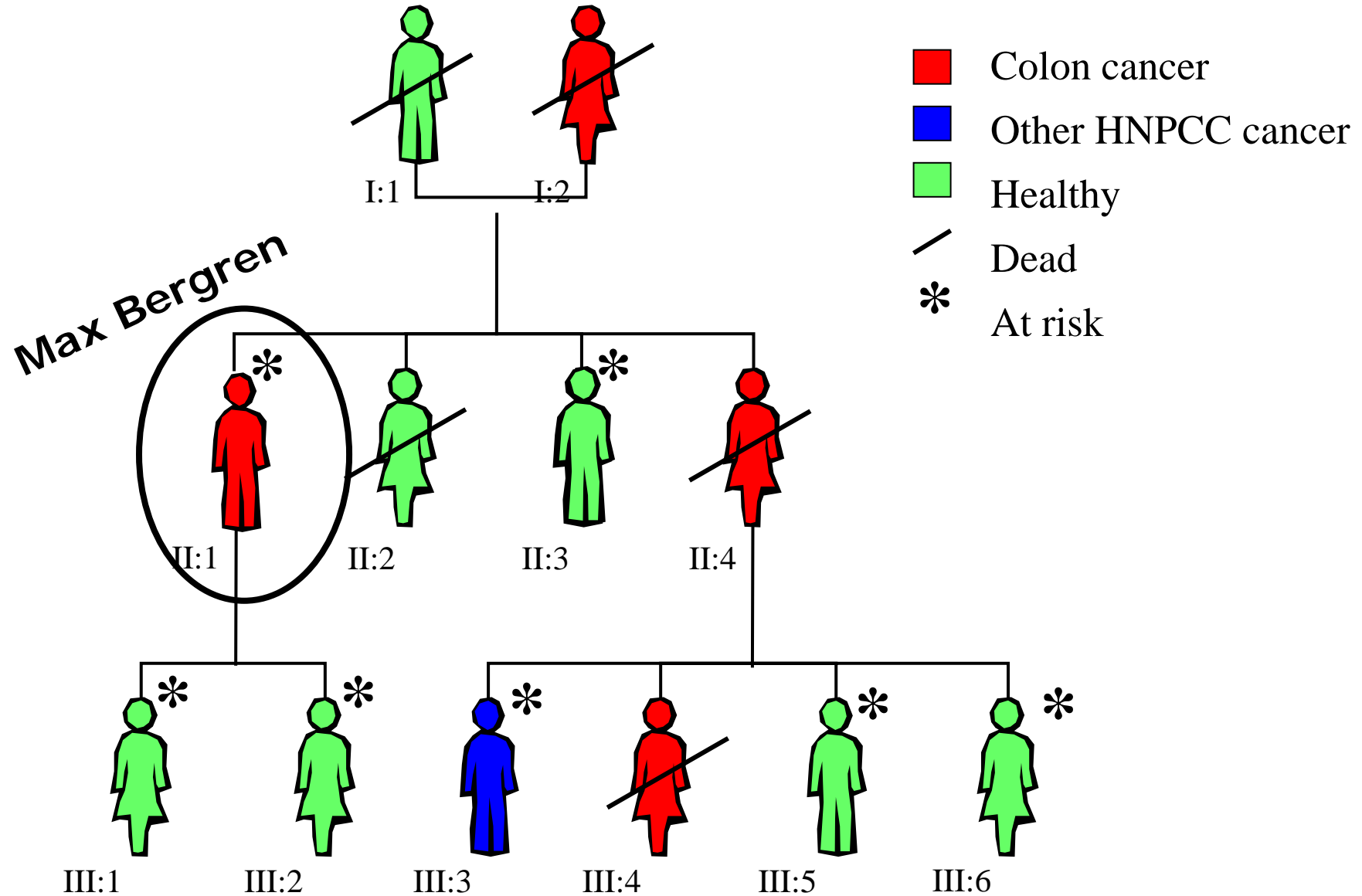
Patient name: **Max Bergren**

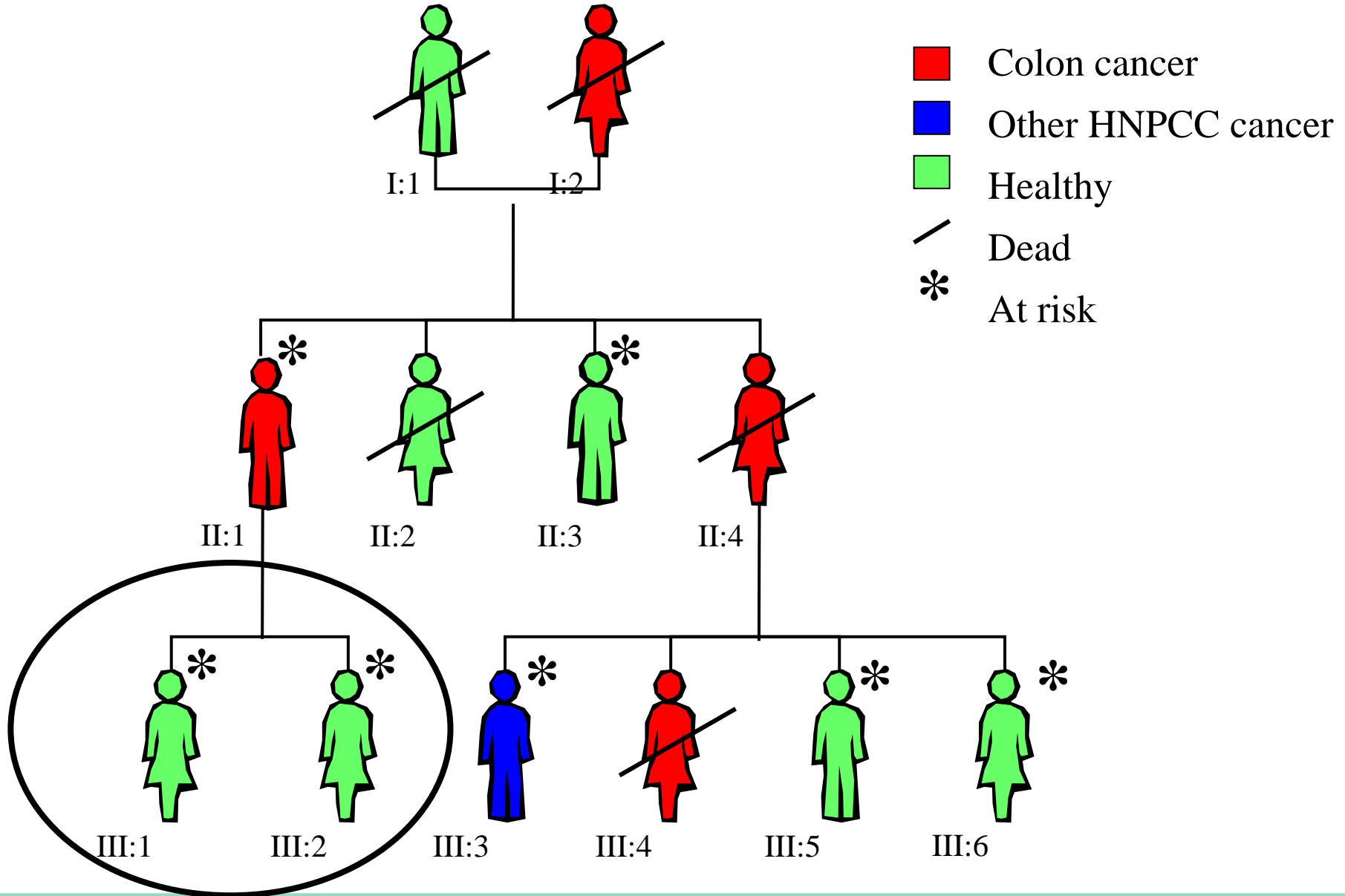
Cpr-nr: **100270-1345**

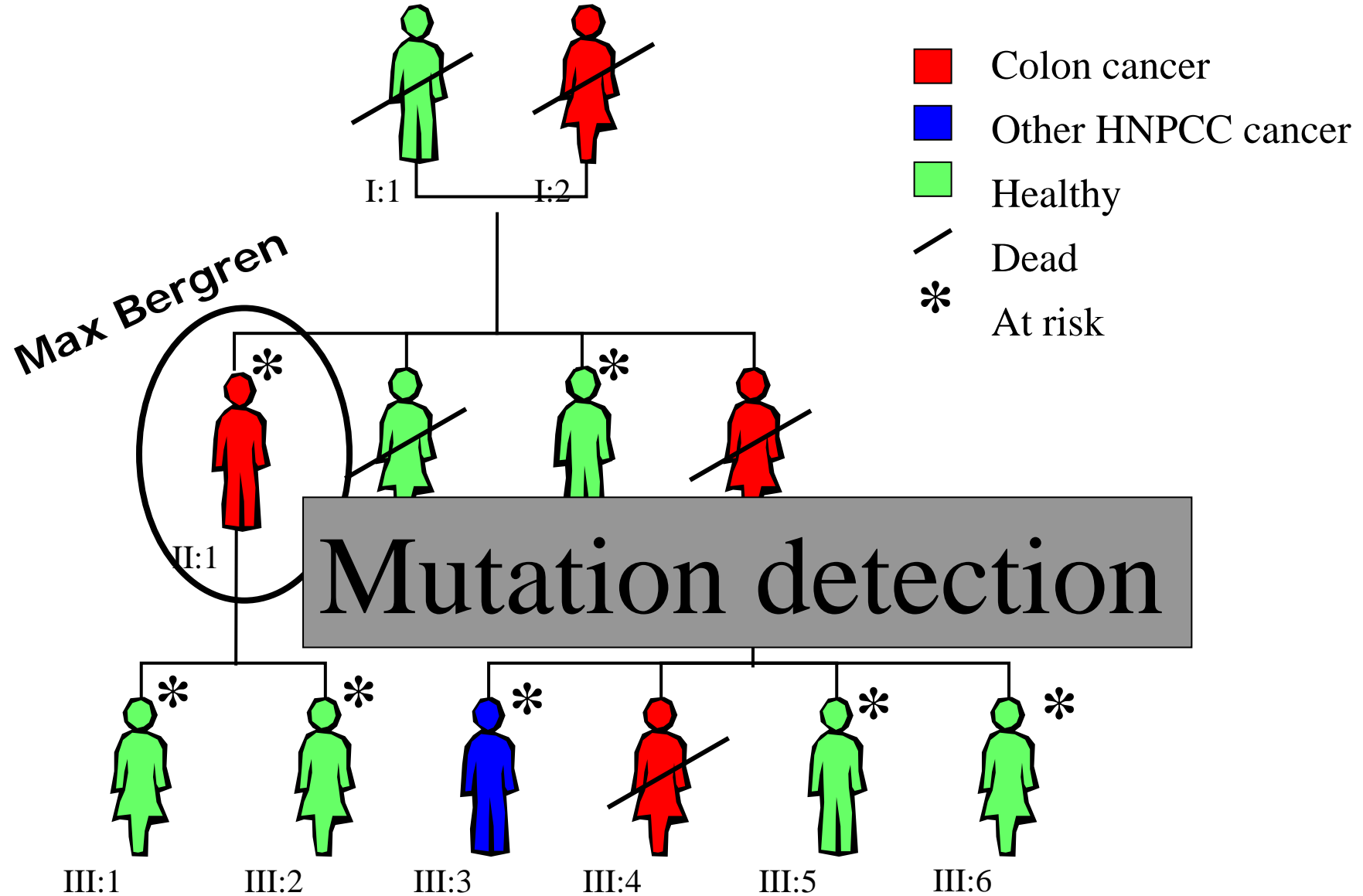
- Patient
- HNPCC
- Treatment
- Results
 - Result
 - Cancers
 - Cancer**
 - Cancer
 - Cancer
 - Result
 - Result
 - Send Report
 - Cancel Report

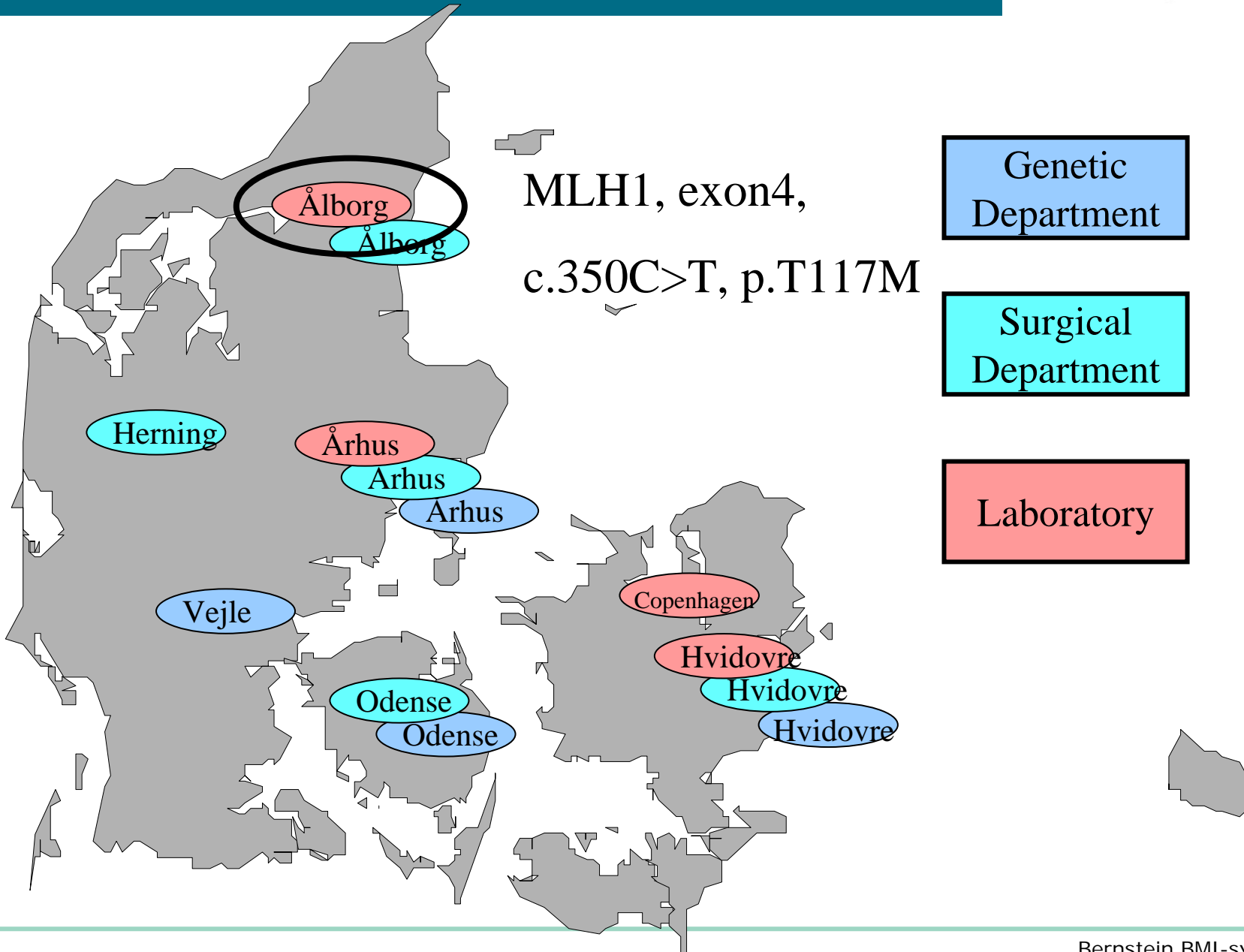
Cancer		 Delete
ICD Code	<input type="text" value="1531"/>	
Description	<input type="text" value="Transverse colon"/>	

HNPCC-registret









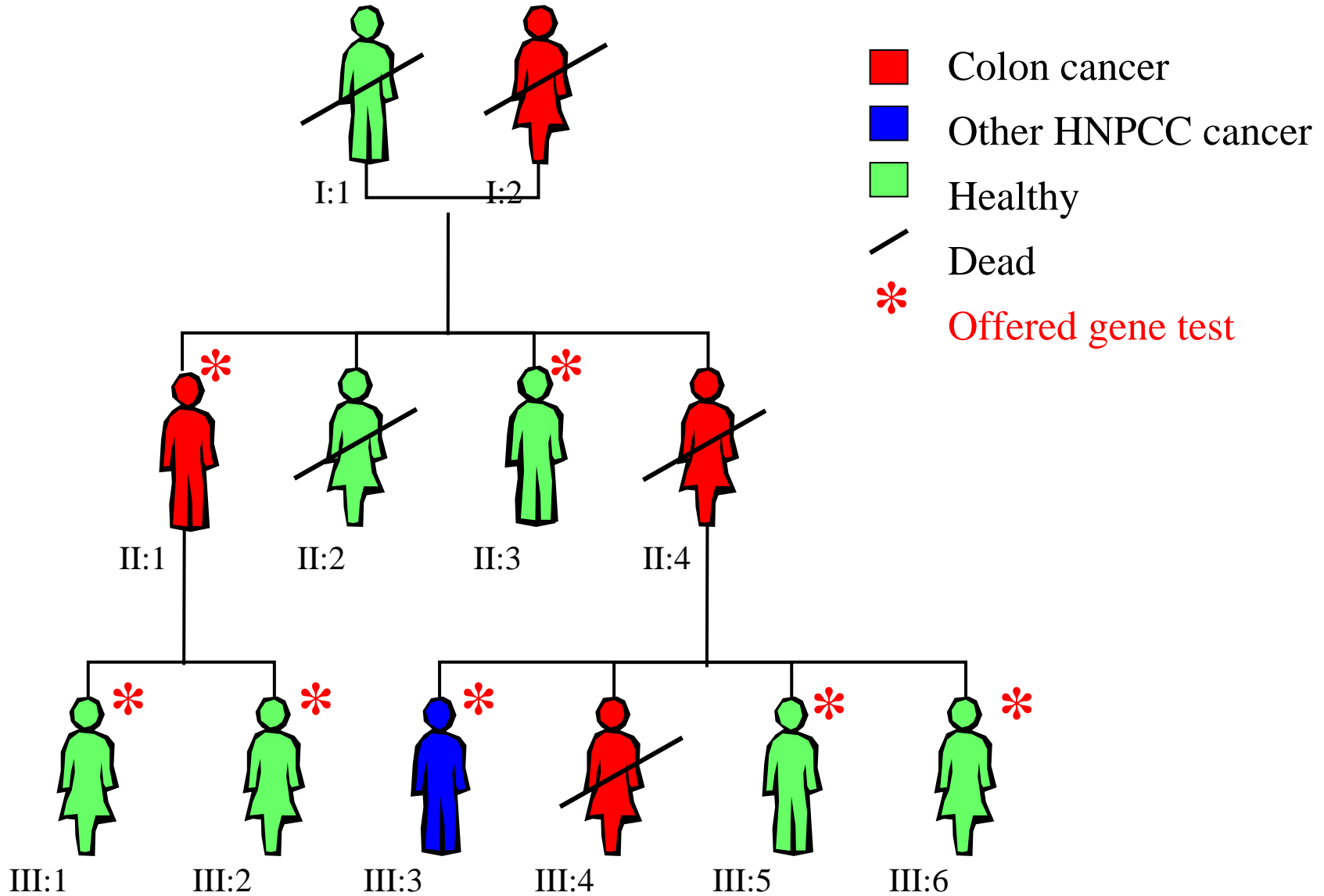
[Report](#) [Statistic](#) [About](#) [Log Out](#)

Report type: **Laboratory Analysis** Patient name: **Max Bergren**
Cpr-nr: **100770-1234**

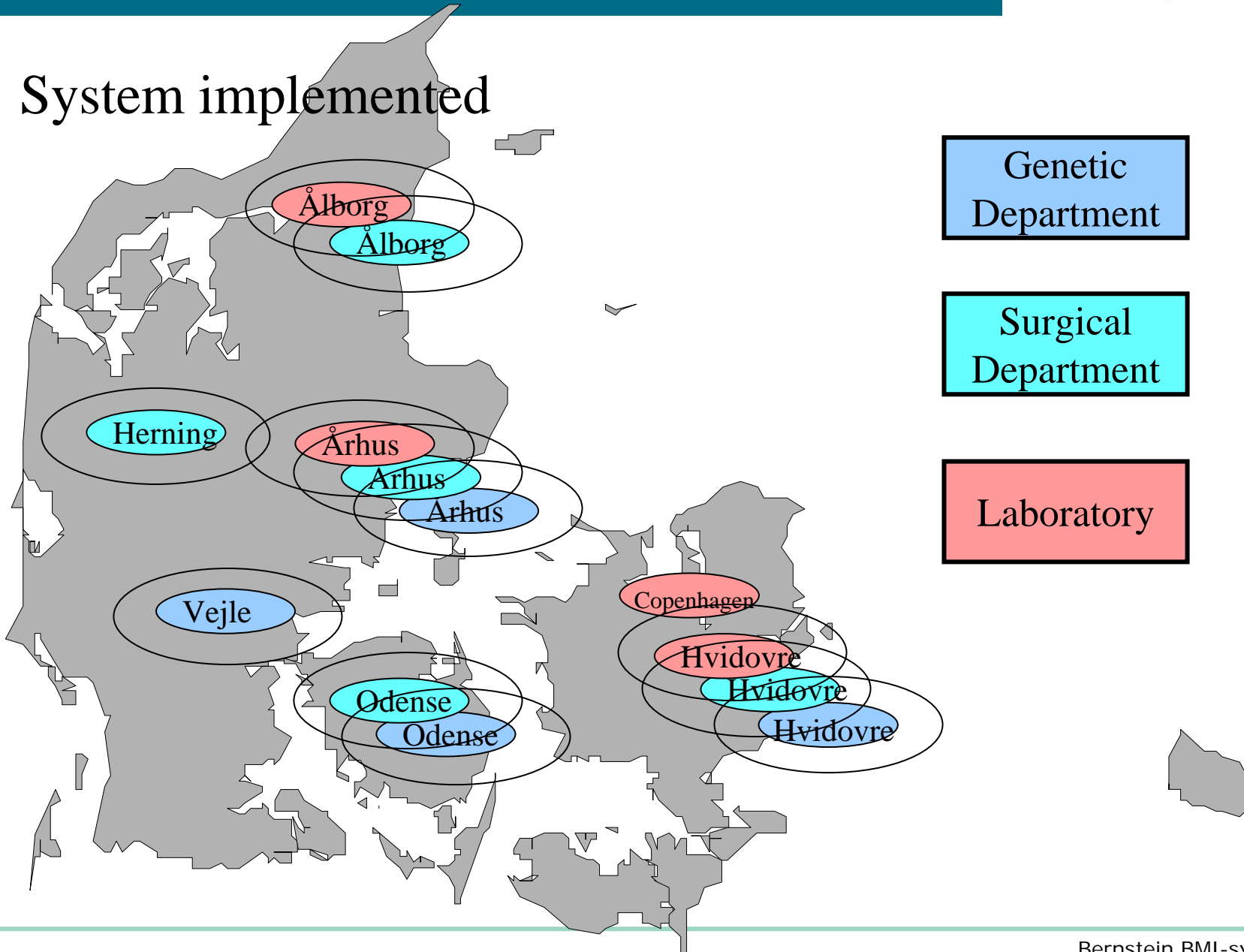
- Patient
- HNPPC
- Gene Codes
 - Gene Code**
 - Variations
- Send Report
- Cancel Report

Gene Code		✕ Delete
Analysis		
Received	<input type="text" value="25-02-06"/>	
Controlled	<input type="text" value="01-03-06"/>	
Answering	<input type="text" value="17-03-06"/>	
Gene Name		
Code	<input type="text" value="1"/>	
Description	<input type="text" value="MLH1"/>	
Method Type		
Code	<input type="text" value="2"/>	
Description	<input type="text" value="DNA-sequencing"/>	
Exons Analysed	<input type="text" value="14"/>	
Protocol		
Code	<input type="text" value="3"/>	
Description	<input type="text" value="MLH1-MDL, Aalborg 12-05-06"/>	

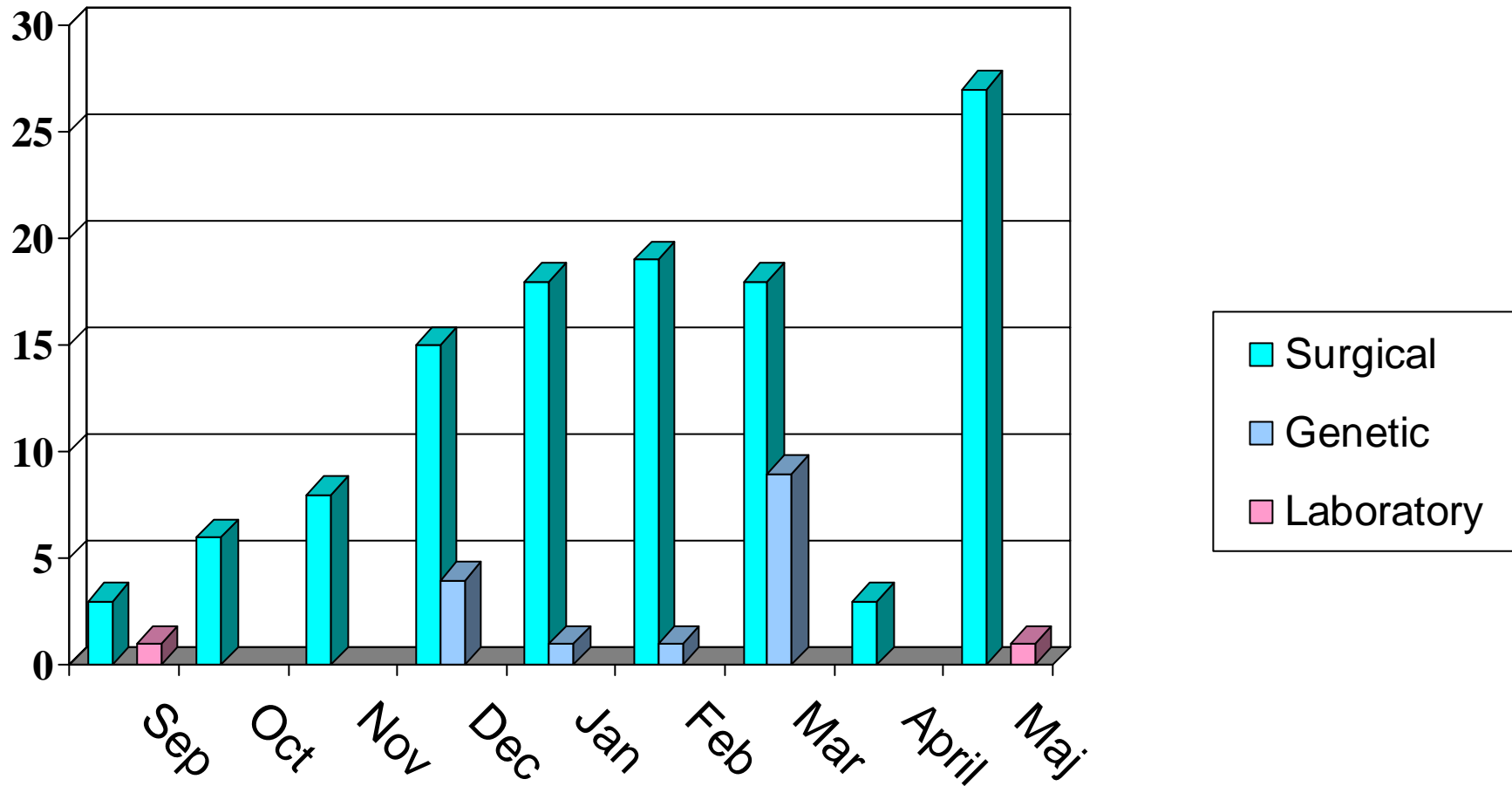
HNPPC-registret



System implemented



134 XML – 87% Surgical dept



WP 6.4 Genomics and colon cancer

Main

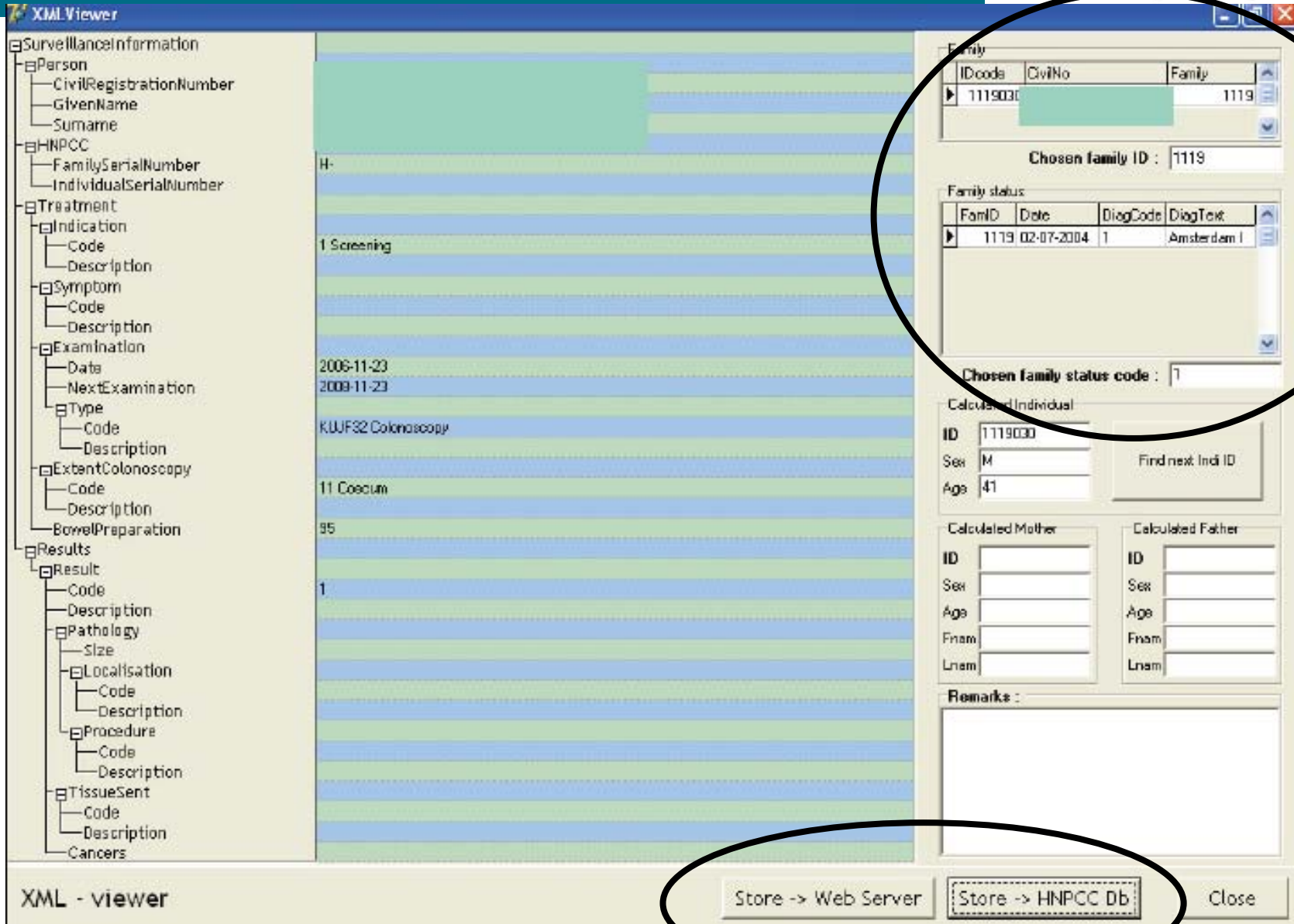
Welcome XML

TimeStamp	FAM	FamilyStatus	INDIVIDUAL_ID	MOTHER_ID	FATHER_ID	Processed	UserName
13-12-2006 10:01:28	1343	4	1343010			False	KIRO3BAEK
13-12-2006 10:05:37						False	KIRO3BAEK
20-12-2006 19:06:26						False	KIRO4RAH
20-12-2006 19:11:58	20000	30	20000002			False	KIRO4RAH
20-12-2006 19:14:34	1622	4	1622076			False	KIRO4RAH
20-12-2006 19:17:16	20000	30	20000004			False	KIRO4RAH
20-12-2006 19:20:11	10000	28	10000002			False	KIRO4RAH
20-12-2006 19:22:34	2017	7	2017010			False	KIRO4RAH
20-12-2006 19:27:24	1632	30	1632014			False	KIRO4RAH
20-12-2006 19:38:05	20000	30	20000006			False	KIRO4RAH
03-01-2007 16:05:25	1546	7	1546010			False	KIRO4RAH
05-01-2007 13:25:11						False	KIRO2LIN
09-01-2007 14:52:56						False	GEN03GER
10-01-2007 20:22:04	813	27	813014			False	KIRO1LAU
10-01-2007 20:25:18	551	4	551122			False	KIRO1LAU
15-01-2007 15:34:22	937	5	937010			False	KIRO2LIN
15-01-2007 15:38:06	1041	22	1041010			False	KIRO2LIN
15-01-2007 15:41:14						False	KIRO2LIN
17-01-2007 11:36:22	489	1	489012			False	KIRO3BAEK
17-01-2007 11:42:17	299	1	299012			False	KIRO3BAEK
17-01-2007 11:45:28	878	22	878020			False	KIRO3BAEK
17-01-2007 11:48:36	1343	4	1343012			False	KIRO3BAEK
24-01-2007 12:03:37	1297	7	1297010			False	KIRO2LIN
26-01-2007 18:59:04	234	3	234034			False	KIRO4RAH
29-01-2007 14:32:14						False	KIRO1LAU
29-01-2007 14:34:08	1279	4	1279036			False	KIRO1LAU
29-01-2007 14:35:58						False	KIRO1LAU
29-01-2007 14:38:11	384	1	384010			False	KIRO1LAU
29-01-2007 14:40:02	1119	1	1119030			False	KIRO1LAU

XML - poster (nye)

Generate reports Edit post

WP 6.4 Genomics and colon cancer



XML Viewer

- Surveillance information
 - Person
 - CivilRegistrationNumber
 - GivenName
 - Surname
 - HNPCC
 - FamilySerialNumber: H-
 - IndividualSerialNumber
 - Treatment
 - Indication
 - Code: 1 Screening
 - Description
 - Symptom
 - Code
 - Description
 - Examination
 - Date: 2006-11-23, 2009-11-23
 - NextExamination
 - Type
 - Code: KWUF32 Colonoscopy
 - Description
 - ExtentColonoscopy
 - Code: 11 Coecum
 - Description
 - BowelPreparation: 95
 - Results
 - Result
 - Code: 1
 - Description
 - Pathology
 - Size
 - Localisation
 - Code
 - Description
 - Procedure
 - Code
 - Description
 - TissueSent
 - Code
 - Description
 - Cancers

IDcode	CiviNo	Family
1119030		1119

Chosen family ID : 1119

FamID	Date	DiagCode	DiagText
1119	02-07-2004	1	Amsterdam I

Chosen family status code : 1

Calculated Individual

ID: 1119030
Sex: M
Age: 41
Find next Indi ID

Calculated Mother

ID:
Sex:
Age:
Fnam:
Lnam:

Calculated Father

ID:
Sex:
Age:
Fnam:
Lnam:

Remarks :

XML - viewer

Store -> Web Server Store -> HNPCC Db Close

Surveillance

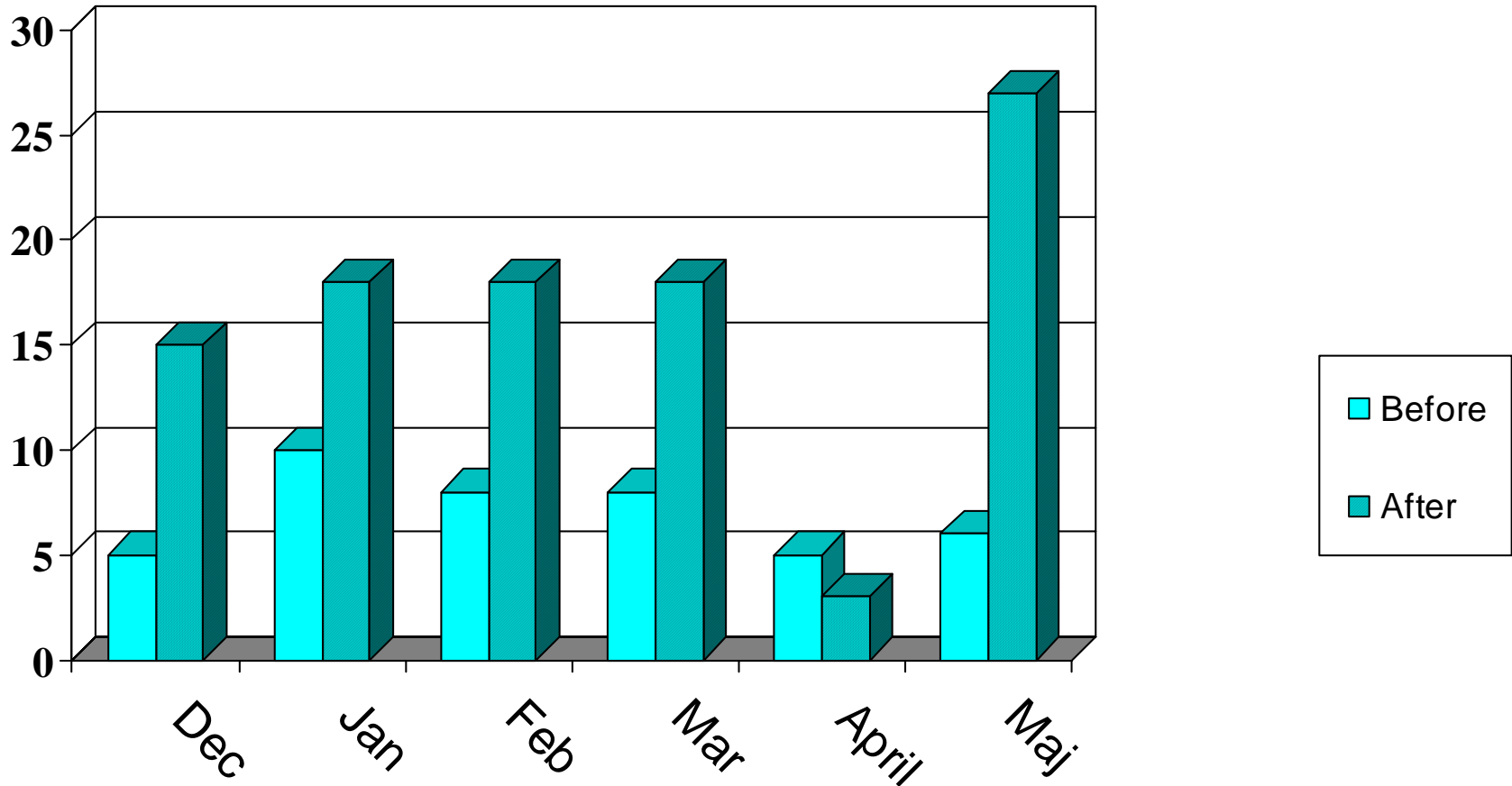
HNPCC-registret

KIR04RAH - Hans Rahr

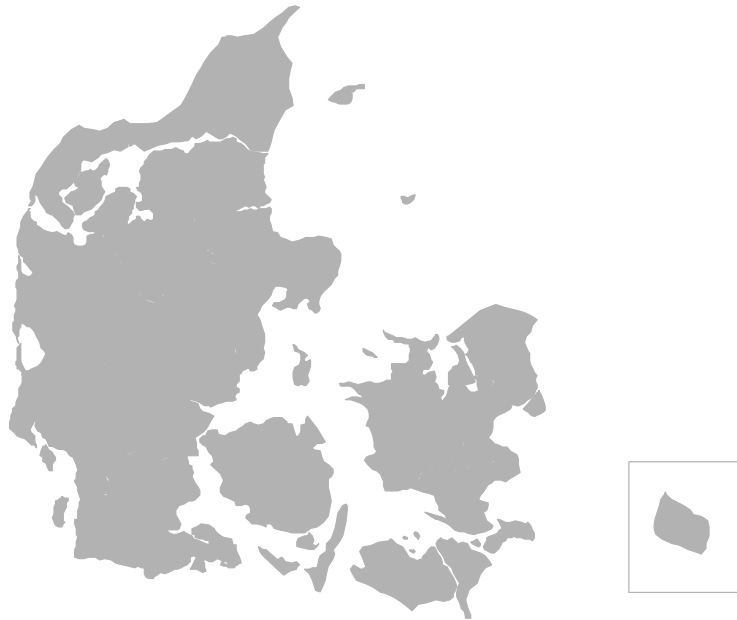
Date 01-02-2007

ID code	Civlino	Family	Family Status	Exam Date	Examination	Result
				2005-10-25	KUIJF32 Colonoscopy	1 Normal
111056		111	3 Amst I+ detected mut	2005-10-25	KUIJF32 Colonoscopy	1 Normal
2060010		2060	27 Late onset suspected	2005-10-07	KUIJF32 Colonoscopy	1 Normal
				2005-12-07	KUIJF32 Colonoscopy	5 Polyp NOS
20000002		20000	30 DCCG-II	2005-12-04	KUIJG01 Proctoscopy + X-ray	1 Normal
1522076		1522	4 Amsterdam I suspected	2005-11-07	KUIJF32 Colonoscopy	1 Normal
20000004		20000	30 DCCG-II	2005-11-16	KUIJF32 Colonoscopy	1 Normal
10000002		10000	28 DCCG-I	2005-11-16	KUIJF32 Colonoscopy	1 Normal
2017010		2017	7 Under evaluation	2005-10-27	KUIJF32 Colonoscopy	1 Normal
1632014		1632	30 DCCG-II	2005-11-26	KUIJF32 Colonoscopy	1 Normal
20000006		20000	30 DCCG-II	2005-11-27	KJFA15 Colonoscopy + polypectomy	5 Polyp NOS 5 Polyp NOS 5 Polyp NOS 5 Polyp NOS
1546010		1546	7 Under evaluation	2005-12-11	KJFA15 Colonoscopy + polypectomy	5 Polyp NOS
234034		234	3 Amst I+ detected mut	2005-10-23	KUIJF32 Colonoscopy	1 Normal

67% increase surveillance reports



Gaps identified



Lack of

Standards

Local databases

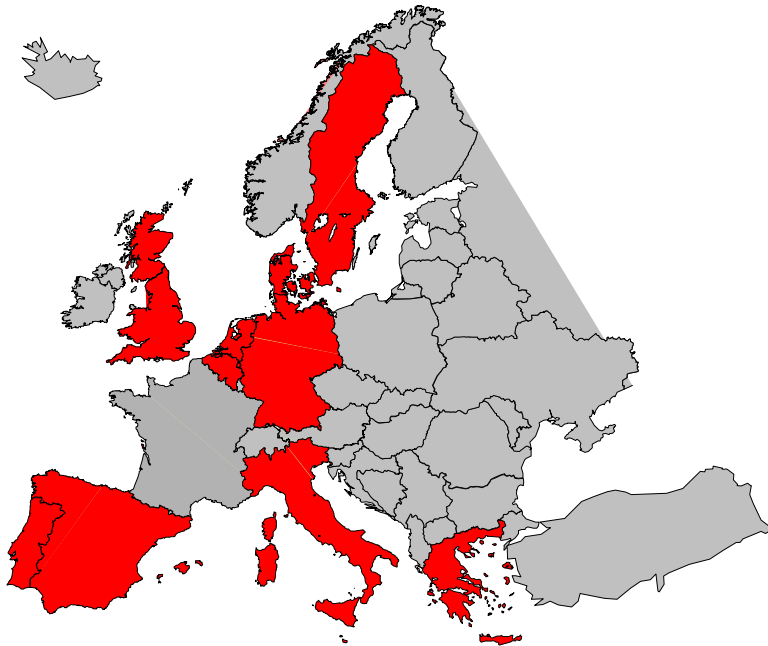
Time-consuming

Implementation

Reporting

Interest – Feed-back

Specific outcome



- Current status data exchange HNPCC
- 4 XML standards on clinical content
- Design of XML documents
- WEB service client
- Experience
 - Implementation
 - Evaluation
 - Dissemination
 - Early HL7 and CDA experience
 - Ethical, legal and security issues

