

Annex 2 - Table of investigated nomenclatures, terminologies and classifications

Legend:

CVD
Diabetes
Oncology
CVD and Diabetes
CVD and Oncology
Diabetes and Oncology
CVD, Diabetes and Oncology

0=No data

1=Data exists

CVD, Diabetes and Breast cancer use case scenario variables	Sub variable	Current data availability in Estonian databases				Data origin (episodes over lifespan)	Current data availability according to standards			
		Estonian Genome Centre	EHIS (e-tervis)	EHIF (Haigekassa)	Hospitals		HL7	LOINC	ATC	Others (e.g. ICD-10, SNOMED, DICOM, GOA, HPO)
Cardiovascular diseases										
Diagnosis		1	1	1	1					ICD-10
Sex		1	1	1	1	Personal ID number				
Weight		1	1	0	1	Birth records, school health form, inpatient case summaries, outpatient case summaries, etc.				SNOMED CT 363809009 or 364589006
Height		1	1	0	1	Birth records, outpatient summary, etc.				SNOMED CT 50373000
Body mass index (BMI)		0	0	0		Calculated				SNOMED CT 60621009
Race		0	0	0						
Family history		1	0	0	0	Genealogy				
	Stroke family history in age less than 65 years	1	with help of external pedigree	with help of external pedigree	with help of external pedigree	Diagnosis				

	Myocardial infarction family history in age less than 65 years	1	1	0	0	Death reasons registry, Inpatient summary			
Laboratory tests	Serum / plasma					Inpatient summary, Outpatient summary, Consultation note			
	S-TSH	0	narrative	0	1			1	EHIF code
	S-T3	0	narrative	0	1			1	EHIF code
	S-T4	0	narrative	0	1			1	EHIF code
	fP-Crea (creatinin plasma level)	0	narrative	0	1			1	EHIF code
	eGFR (estimated glomerular filtration rate)	0	narrative	0	1			1	EHIF code
	S-Gluc (blood glucose)	0	narrative	0	1			1	EHIF code
	HbA1c (Hemoglobin A1c)	0	narrative	0	1			1	EHIF code
	S-Chol (total cholesterol)	0	narrative	0	1			1	EHIF code
	HDL-cholesterol	0	narrative	0	1			1	EHIF code
	LDL-cholesterol	0	narrative	0	1			1	EHIF code
	Triglycerides	0	narrative	0	1			1	EHIF code
	Uric acid	0	narrative	0	1			1	EHIF code
	Urine								
	Albumin/creatinin ratio in urine	0	narrative	0	1			1	EHIF code

ECG	Left ventricular hypertrophy	0	narrative	0	narrative				
Echocardiography	Left ventricular hypertrophy	0	narrative	0	narrative				
Radiology exam									
	Ultrasound of kidneys (images)	0	0	0	1				Estonian nation-wide PACS (picture archiving and communication system)
	Ultrasound of kidneys (report)	0	narrative	0	narrative				Estonian nation-wide PACS
Smoking		1	1	0	1	Health check notes, Dental check notes			SNOMED CT 405140009
Dyslipidaemia	S-Chol (total cholesterol)	0	narrative	0	1			1	EHIF code
	HDL-cholesterol	0	narrative	0	1			1	EHIF code
	LDL-cholesterol	0	narrative	0	1			1	EHIF code
	Triglycerides	0	narrative	0	1			1	EHIF code
Metabolic syndrome						Diagnose?			ICD10
Pre-treatment pulse rate		1	1	0	1	Outpatient summary, Health check notes			SNOMED CT 364075005
On-treatment pulse rate		1	1	0	1	Outpatient summary, Health check notes			SNOMED CT 364075005

Pre-treatment blood pressure										
	Systolic	1	1	0	1	Outpatient summary, Health check note				SNOMED CT 271649006
	Diastolic	1	1	0	1	Outpatient summary, Health check note				SNOMED CT 271650006
Pre-treatment ambulatory blood pressure monitoring (ABPM)		0	0	0						
	mean 24-hour		narrative	1	1		1			
	daytime systolic blood pressure		narrative	1	1		1			
	daytime diastolic blood pressure		narrative	1	1		1			
	night-time systolic blood pressure		narrative	1	1		1			
	night-time diastolic blood pressure		narrative	1	1		1			
	Nocturnal blood pressure fall (%)		narrative	1	1		1			
	mean 24-hour heart rate		narrative	1	1		1			
On-treatment blood pressure readings										
	Systolic									
	Diastolic									

On-treatment ambulatory blood pressure monitoring (ABPM)									
	mean 24-hour		narrative	1	1				
	daytime systolic blood pressure		narrative	1	1				
	daytime diastolic blood pressure		narrative	1	1				
	night-time systolic blood pressure		narrative	1	1				
	night-time diastolic blood pressure		narrative	1	1				
	Nocturnal blood pressure fall (%)		narrative	1	1				
	mean 24-hour heart rate		narrative	1	1				
	Blood pressure control according to office blood pressure measurements		narrative	1	1				
	Blood pressure control according to home blood pressure measurements		narrative	1	1				
	Blood pressure control according to ABPM		narrative	1	1				
Medication									
	Prescribed medication	declared	1	1	1	Prescription, Dental care notes, Inpatient summary,			1

						Outpatient summary			
	Over the counter medication	declared	narrative	0	narrative				
Compliance to medication	According to electronic prescription database (number and proportion of purchased antihypertensive drugs during last 6 months)	0	0	1	0	Prescription dispensing			1
Lifestyle modification									
	Body weight lowering								Continua alliance
	Decrease in salt consumption								
Number of general practitioner visits / consultations	During last 12 months	0	1	1	0	Outpatient summary, Reimbursement claim			
Number of specialists visits / consultations	During last 12 months	0	1	1	0	Outpatient summary, Consultation notes, Reimbursement claim			
Reminders to the patient									

	Obesity (E66.0), Hypertension (I10),Chronic urinary tract infection (N39.0	1	1	1	1	Diagnose				ICD10
Diabetes Risk Questionnaire			narrative							
Genotype score based on diabetes risk alleles which may predict new cases		0	0	0	0					
First diagnosis of diabetes		1	1	1	1	Diagnose				ICD10
Glucose tolerance test			narrative				1			
Long term complications	Diagnoses	1	1	1	1	Diagnose				ICD10
	Procedures	0	1	1	1	Procedure				NCSP
Number of visits to diabetes nurse		0	1	1	0	Nursing notes, Outpatient summary, Reimbursement claims				
Number of visits to GP nurse		0	1	1	0	Nursing notes, Outpatient summary, Reimbursement claims				
Medication										
	Prescribed oral medication			1		Prescriptions			1	

	Prescribed insulin injections			1		Prescriptions			1
	Over the counter medication								
	Concomitant medications – statins, hypertension treatment			1					1
	Test stripes, glucometers, needles for pen-syringes and lancets, etc.			1		Medical card dispensing			
Ophthalmological examination of eye fundus or photography of eye fundus			narrative						
Peripheral pulses on leg (Doppler of arteries on leg)			narrative						
Neurological sensitivity investigation			narrative						
	Physical examination		narrative						
	ENMG (Electroneuromyography)		narrative						
Photos of diabetic leg ulcers									Estonian nation-wide PACS
Diabetes diary									Continua alliance
Continues glucose monitoring with sensor									Continua alliance

	status								
	HER2 status	narrative							
Radiology exam	Mammography (report)	1	1						
	Mammography (images)								Estonian nation-wide PACS
	Ultrasonography: breasts (report)	1							
	Ultrasonography: breasts (images)								Estonian nation-wide PACS
	Ultrasonography: abdomen, pelvis (report)	1							
	Ultrasonography: abdomen, pelvis (images)								Estonian nation-wide PACS
	X-ray: thorax (report)	1							
	X-ray: thorax (images)								Estonian nation-wide PACS
	MRT breasts (report)	1							
	MRT breasts (images)								Estonian nation-wide PACS
	MRT: (report)	1							
	MRT: (images)								Estonian nation-wide PACS
	MRT: abdomen, pelvis (report)	1							
	MRT: abdomen, pelvis (images)								Estonian nation-wide PACS
	CT brain (report)	1							
	CT brain (images)								Estonian nation-wide PACS

	CT thorax (report)		1							
	CT thorax (images)									Estonian nation-wide PACS
	CT abdomen (report)		1							
	CT abdomen (images)									Estonian nation-wide PACS
	CT pelvis (report)		1							
	CT pelvis (images)									Estonian nation-wide PACS
	PET (report)		1							
	PET (images)									Estonian nation-wide PACS
	SPECT-CT (report)		1							
	SPECT-CT (images)									Estonian nation-wide PACS
Therapy	Type of surgery									NCSP
	Medical therapies (drugs, regimens, continuations of therapies)				1					1
	Type of neoadjuvant chemotherapy and biological therapy		narrative							
	Type of adjuvant chemotherapy and biological therapy		narrative							
	Type of adjuvant hormonal therapy		narrative							
	Type of metastatic chemo- and biological therapy		narrative							

	Type of metastatic hormonal therapy		narrative							
	Type of radiotherapy		narrative							
Genes Analysed in Commercially Available Multiplex Panels										
	APC									
	ATM									
	ATR									
	BABAM1									
	BAP1									
	BARD1									
	BMPR1A									
	BRIP1									
	CDH1									
	CDK4									
	CDKN2A									
	CHEK1									
	CHEK2									
	FAM175A/Abraxas									
	MLH1									
	MRE11A									
	MSH2-positive EPCAM									
	MSH6									
	MUTYH									
	NBN									
	PALB2									

	PMS2									
	PRSS1									
	PTEN									
	RAD50									
	RAD51									
	RAD51B									
	RAD51C									
	RAD51D									
	RBBP8									
	RET									
	SMAD4									
	STK11									
	TP53									
	TP53BP1									
	UIMC1									
	VHL									
	XRCC2									
	XRCC3									