Kühtai 14. April 2023

Detailed Clinical Phenotyping in Precision Medicine

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Acknowledgments

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Iterata AG, Gränichen, Switzerland Dr. Hansjörg Lehner Stefan Hubeli Urs Rutschi

Aims

To confer basic knowledge of the description of physical characteristics of patients

To describe the current state of systems for defining, categorizing and capture of these characteristics

To explain the problems faced when implementing a clinically usable data capture system and to provide a working

Introduction

Which data of phenotyping to rely on?

- How to capture and store clinical data?
- How to enable contextual assignments, categorization and analytic evaluation of the data
- How to use the data to answer clinically decisive questions for achieving optimal outcomes?

What is a phenotype? – Peter Robinson, Charité, Berlin

- At least five definitions of phenotype in use in the biology literature
- The collection of observable traits of an organism, comprising its morphology, its physiology at the level of the cell, the organ, and the body, and its behaviour, comprising even characteristics such as the gene expression profiles in response to environmental cues
 - Robinson P Human Mutation 2012;33:778

Phenotype for purely clinical purposes

- Collection of traits elicited from the health history and physical findings of a given patient
- **Precision medicine**
 - Using the comprehensive phenotype and all available, validated medical knowledge to tailor patient care with the aim of achieving an optimal outcome

Systems for Clinical Phenotyping

Clinical data from diagnostic classification systems of diseases

Standardized nomenclature systems

- SNOMED
- ICD (International Classification of Diseases)
- ICF (International Classification of Functioning)

DECIPHER (DatabasE of genomiC variation and Phenotype in Humans using Ensembl Resources)

Human Phenotype Ontology (HPO)

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About	Statistics	News archiv	e Citing	Data sharing	Downloads	Legal	Cookies policy	Advisory board	Affiliations		Help	FAQs

Background

Many patients suffering from Rare Disease harbour genomic variants (sequence variants or copy number variants) that by disrupting normal gene expression lead to disease. However, many variants are novel or extremely rare, making clinical interpretation problematic and genotype-phenotype correlations uncertain. Identification of patients sharing variants in a given gene and having phenotypic features in common leads to greater certainty in the pathogenic nature of the gene and enables to the role of novel genes in development and disease to be defined. Furthermore, analysis of the type of genomic variant and of its consequence (eg. Loss of function or gain of function) enables insight into the mechanism of disease and potential therapeutic targets.

DECIPHER Project Proposal 🕹

DECIPHER

DECIPHER (DatabasE of genomiC variation and Phenotype in Humans using Ensembl Resources) is an interactive web-based database which incorporates a suite of tools designed to aid the interpretation of genomic variants.

DECIPHER enhances clinical diagnosis by retrieving information from a variety of bioinformatics resources relevant to the variant found in the patient. The patient's variant is displayed in the context of both normal variation and pathogenic variation reported at that locus thereby facilitating interpretation.

The DECIPHER Community

Contributing to the DECIPHER database is an international community of academic departments of clinical genetics and rare disease genomics now numbering more than 270 centres and having uploaded more than 36,000 cases. Each contributing centre has a nominated rare disease clinician or clinical geneticist who is responsible for overseeing data entry and membership for their centre. DECIPHER enables a flexible approach to data-sharing. Each centre maintains control of its own patient data (which are password protected within the centre's own DECIPHER project) until consent is given to share the data with chosen parties in a collaborative group or to allow anonymous genomic and phenotypic data to become freely viewable within Ensembl and other genome browsers (see below). Once data are shared, consortium members are able to gain access to the patient report and contact each other to discuss patients of mutual interest.

Public Data Access

With patient consent, positional genomic information together with a brief description of the associated phenotype becomes viewable without password protection, for example, via the DECIPHER track in Ensembl. This is of benefit not only to clinicians advising patients with similar findings but also to researchers working on specific phenotypes, Rare Diseases, drug targets or the role of genes in health and development.

https://www.deciphergenomics.org

DECIPHER (DatabasE of genomiC variation and Phenotype in Humans using Ensembl Resources)

- Interactive web-based database which incorporates a suite of tools designed to aid the interpretation of genomic variants
- DECIPHER enhances clinical diagnosis by retrieving information from a variety of bioinformatics resources relevant to the variant found in the patient
- Patient's variant displayed in the context of both normal variation and pathogenic variation reported at that locus, thereby facilitating interpretation

https://www.deciphergenomics.org



(a) DECIPHER enables the deposition of phenotypes using HPO terms. (b) DECIPHER supports the deposition of developmental milestones and anthropometric measurements, for example, occipitofrontal (head) circumference

Foreman J Human Mutation 2022;43:682-697



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Phenotypic abnormality in open-access patients in DECIPHER

Hide redundant paths

Simple view About



Abnormality of the cardiovascular system Abnormality of blood and blood-forming tissues Abnormality of the endocrine system Abnormality of the musculoskeletal system Abnormality of the digestive system Abnormality of limbs Abnormality of prenatal development or birth Growth abnormality Abnormality of the genitourinary system Abnormality of the ear Abnormality of metabolism/homeostasis Abnormal cellular phenotype Abnormality of the eye Abnormality of the nervous system Neoplasm Constitutional symptom Abnormality of the immune system Abnormality of the voice Abnormality of the respiratory system Abnormality of head or neck Abnormality of the breast Abnormality of the integument



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Phenotypic abnormality in open-access patients in DECIPHER



Search results for "HP:0010978" (Refine Search)



Filter...

Patients: 11 to 20 of 1417

DECIPHER Patient ID	Sex	Phenotype(s)	Variants	Contact
•	€ ≑	*	¢	
1375	46XY	Accessory oral frenulum, Alveolar ridge overgrowth, Delayed cranial suture closure, Diastasis recti, Feeding difficulties in infancy, Frontal bossing, Generalized hirsutism, High palate, Hoarse voice, Hyperactivity, Hypotonia, Intellectual disability, Low-set ears, Midface retrusion, Pes planus, Recurrent infections, Ridged cranial sutures, Strabismus	1	
1593	46XX	Abnormal heart morphology, Abnormal pinna morphology, Abnormality of the tarsal bones, Abnormality of the upper respiratory tract, Blepharophimosis, Blue sclerae, Feeding difficulties in infancy, Hypertelorism, Hypotonia, Intellectual disability, Microcephaly, Micrognathia, Patent ductus arteriosus, Recurrent infections, Seizure, Short stature, Single transverse palmar crease, Sleep disturbance, Wide nasal bridge	4	
1721	Other	Atopic dermatitis, Ichthyosis	3	
1925	46XY	Abnormal pinna morphology, Dolichocephaly, Exaggerated cupid's bow, Hypermetropia, Intellectual disability, Posteriorly rotated ears, Recurrent infections	1	
1993	Other	Cleft of alveolar ridge of maxilla, Constipation, Hypoglycemia, Lipodystrophy, Recurrent infections, Small nail, Sparse hair	2	
1995	Other	Abnormal hair pattern, Abnormality of the forehead, Arachnodactyly, Constipation, Multiple renal cysts, Recurrent infections, Tapered finger	3	
2068	46XX	Atopic dermatitis, Clinodactyly of the 5th finger, Delayed speech and language development, Fine hair, Intellectual disability, Microcephaly, Prominent nose, Short hallux, Short nail, Short palm, Short thumb, Tetralogy of Fallot, Tooth malposition	3	
2069	46XX	Atopic dermatitis, Autism, Behavioral abnormality, Bilateral tonic-clonic seizure, Brachycephaly, Constipation, Delayed speech and language development, Echolalia, Hypertelorism, Intellectual disability, Low anterior hairline, Strabismus, Widely spaced teeth	1	
2126	46XX	Abnormality of the periorbital region, Ataxia, Constipation, Delayed speech and language development, Intellectual disability, Microcephaly, Recurrent infections, Recurrent urinary tract infections, Short stature, Stridor	2	
2154	46XX	Abnormal dental enamel morphology, Atopic dermatitis, Bulbous nose, Congenital diaphragmatic hernia, Feeding difficulties in infancy, Hypermetropia, Hypotonia, Intellectual disability, Macrotia, Microcephaly, Scoliosis, Small for gestational age, Thin upper lip vermilion	1	
10	*	Previous 1 2 3 4 5 142 Next		



e.g. Arachnodactyly | Marfan syndrome | FBN1

The Human Phenotype Ontology

About

The Human Phenotype Ontology (HPO) provides a standardized vocabulary of phenotypic abnormalities encountered in human disease. Each term in the HPO describes a phenotypic abnormality, such as Atrial septal defect. The HPO is currently being developed using the medical literature, Orphanet, DECIPHER, and OMIM. HPO currently contains over 13,000 terms and over 156,000 annotations to hereditary diseases. The HPO project and others have developed software for phenotype-driven differential diagnostics, genomic diagnostics, and translational research. The HPO is a flagship product of the Monarch Initiative, an NIH-supported international consortium dedicated to semantic integration of biomedical and model organism data with the ultimate goal of improving biomedical research. The HPO, as a part of the Monarch Initiative, is a central component of one of the 13 driver projects in the Global Alliance for Genomics and Health (GA4GH) strategic roadmap.

News & Updates

April 2023 HPO release & updates	April 6, 2023
June 2022 HPO release	June 12, 2022
HPOA release	April 15, 2022

earn More About HPO

Exomiser Evaluate variants based on the predicted pathogenicity.

Genomiser Analyze genome sequence data for non-coding variants.



Phenomizer Rank disease differential diagnosis by clinical features.

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Profile Search Discover diseases with a phenotype profile.



Phenopackets

A Global Alliance for Genomics and Health (GA4GH) international standard for phenotypic data exchange.

https://hpo.jax.org/app/

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Human phenotype ontology

Standardized vocabulary of phenotypic abnormalities in human disease

In development using

- Medical literature
- Orphanet (https://www.orpha.net/consor/cgibin/index.php)
- DECIPHER (https://www.deciphergenomics.org)
- OMIM (Online Mendelian Inheritance in Man; https://www.omim.org)

HPO currently contains over 13,000 terms and over 156,000 annotations to hereditary diseases

About



e.g. Arachnodactyly | Marfan syndrome | FBN1

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A Global Alliance for Genomics and Health (GA4GH) international standard for phenotypic data exchange.

https://hpo.jax.org/app/

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Phenomizer

April 2023

- 11'442 features
- 8'057 diseases

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HP:0001459	algorithm for ranking the differential diagnoses.											
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HP:0006088	Köhler et al., Clinical diagnostics in human genetics with se	amantic										
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HP:0010713	Köhler et al. The Human Phenotyne Ontology in 2017											
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HP:0000280	Coarse facial features	0.
HP:0000405	Conductive hearing impairment	0.
HP:0006180	Crowded carpal bones	0.
HP:0000028	Cryptorchidism	0.
HP:0000689	Dental malocclusion	0.
HP:0003083	Dislocated radial head	0.
HP:0000494	Downslanted palpebral fissures	0.
HP:0005463	Elongated sella turcica	0.
HP:0000286	Epicanthus	0.
HP:0001508	Failure to thrive	0.
HP:0000293	Full cheeks	0.
HP:0002857	Genu valgum	0.
HP:0000218	High palate	0.
HP:0001007	Hirsutism	0.
HP:0000238	Hydrocephalus	0.
HP:0000047	Hypospadias	0.
HP:0000023	Inguinal hernia	0.
HP:0002566	Intestinal malrotation	0.

Alg										
	p-value. 🔺	Disease Id.	Disease name.	Genes.						
7	0.3926	OMIM:60	#602080 PAGET DISEASE OF BONE; PDB							
7	0.3926	OMIM:16	#166600 OSTEOPETROSIS, AUTOSOMAL DOMINANT 2; OPTA2;;OSTEOPETROSIS, AUTOSOMAL DOM	CLCN7 (1186)						
7	0.3926	OMIM:61	#613390 FANCONI ANEMIA, COMPLEMENTATION GROUP O; FANCO	FANCC (2176						
7	0.3926	OMIM:60	GRANGE SYNDROME							
7	0.3926	OMIM:61	#615066 OSTEOGENESIS IMPERFECTA, TYPE XIV; OI14 ;;OI, TYPE XIV	SPARC (6678						
7	0.3926	ORPHAN	IDIOPATHIC JUVENILE OSTEOPOROSIS	WNT3A (897						
7	0.3926	ORPHAN	OSTEOGENESIS IMPERFECTA - RETINOPATHY - SEIZURES - INTELLECTUAL DISABILITY							
7	0.3926	OMIM:12	CYSTIC ANGIOMATOSIS OF BONE, DIFFUSE							
	0.4433	OMIM:27	SPINAL MUSCULAR ATROPHY, TYPE I, WITH CONGENITAL BONE FRACTURES							
	0.4838	OMIM:24	HYPOPROTEINEMIA, HYPERCATABOLIC	B2M (567)						
	0.4838	OMIM:60	RADIOULNAR SYNOSTOSIS WITH AMEGAKARYOCYTIC THROMBOCYTOPENIA	HOXA11 (3207						
	0.4838	OMIM:61	#614900 DIAMOND-BLACKFAN ANEMIA 11; DBA11	TSR2 (90121						
	0.4838	OMIM:10	ARMS, MALFORMATION OF							
	0.4838	OMIM:15	MESOMELIC DYSPLASIA, KANTAPUTRA TYPE							
	0.4838	OMIM:21	#212780 CENANI-LENZ SYNDACTYLY SYNDROME; CLSS;;CENANI SYNDACTYLISM;;CENANI-LENZ SY	APC (324), L						
	0.4838	OMIM:17	RADIAL HYPOPLASIA, TRIPHALANGEAL THUMBS, HYPOSPADIAS, AND MAXILLARYDIASTEMA							
	0.4838	OMIM:17	RADIAL RAY HYPOPLASIA WITH CHOANAL ATRESIA							
	0.4838	OMIM:61	#613951 FANCONI ANEMIA, COMPLEMENTATION GROUP P; FANCP	FANCC (2176						
	0.4838	OMIM:12	127350 DYSCHONDROSTEOSIS AND NEPHRITIS							
	0.4838	DECIPHE	LERI-WEILL DYSCHONDROSTOSIS (LWD) - SHOX DELETION							
	0.4838	OMIM:15	%156230 MESOMELIC DWARFISM OF HYPOPLASTIC TIBIA AND RADIUS TYPE							
	0.4838	OMIM:19	ULNAR HYPOPLASIA							
	0.4838	OMIM:11	113470 BRACHYMESOMELIA-RENAL SYNDROME							
	0.4838	OMIM:22	FACIOCARDIOMELIC DYSPLASIA, LETHAL							
	0.4838	OMIM:61	SKELETAL DEFECTS, GENITAL HYPOPLASIA, AND MENTAL RETARDATION	ZBTB16 (7704)						
	0.4838	OMIM:31	ULNAR HYPOPLASIA WITH LOBSTER-CLAW DEFORMITY OF FEET							
	0.4838	OMIM:60	VENTRICULOMEGALY WITH DEFECTS OF THE RADIUS AND KIDNEY							

The Phenomizer

Impediments to use in clinical medicine

Documentation of health history and physical findings

- Insufficient resolution
- No system for documentation or reporting

Features mostly related to genetic defects Incomplete ontology due to

- Lacking resolution
- Binary and specific search aids insufficient
- Omission of standard diagnostic attributes

Multiple time points not possible

Curation centralized Changes require extensive resources Language restricted to English

Data capture for clinical medicine

How to capture and store clinical data?

How to enable contextual assignments, categorization and analytic evaluation of the data

How to operationalize our data to ask and answer decisive questions about outcomes?

Prerequisites

- 1. Unrestricted ability to migrate
 - Data (-base)
 - Programmes/ algorithms
- 2. No proprietary storage system
- 3. Full addressability of data at blinding speed (search engine technology)

Concept

To provide a **comprehensive basis for digital diagnostics**:

- Structured data entry (SDE) tool to capture clinical data as discrete items/features with references to defined properties, anatomic location and time
- Application of advanced IT tools to consolidate and analyse data from different sources to enable evaluation of large cohorts

To enable seamless integration of existing genomic and imaging data of large cohorts of patients with all other modalities* in an easy to use clinical decision support system (CDSS):

- By directly targeting the data stream of highthroughput, allelic level resolution whole genome sequencing (WGS)
- By examining the prospect for image analysis in addition to routinely collected (unstructured) data in clinical care
- By aggregating data to obtain data-driven diagnostic and prognostic information to support precise and fast clinical decision making and personalised medicine

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Documentation of a health history and physical findings

- Efficient and reliable electronic capture
- Time-stamped, discrete data objects and their attributes
- Incorporated real-time report generator
- Multiple language data entry and report
- Data storage system provides basis for broad analysis and inter-actability of data with other storage

The user interface is accessible in two roles, a consumer role (CR) and an administrator role (AR); designated domain experts get access in AR

In AR the domain expert has access to backend subsystems that permit

- To create, modify and delete fields
- To set field-properties, such as the type (text field, checkbox, radio-button, etc.)
- To position
- To create new forms

Content and phrasing of the report generator can be defined in AR

Clinicians use the CR to create new content of existing forms, alter content, search for content, and generate reports

Captured content is processed in a semantically cohesive format by real-time report generation with the intention to spare physician and allied health care personnel resources

Allgemein

Haut und Nägel

Kopf und Hals

Lymphknoten

Thorax

Kreislauf

Abdomen

Neurostatus

Bewegungsapparat

Allgemeinzustand 🖸 gut pathologisch Details Bewusstsein normal pathologisch somnolent soporös auf Schmerzreiz reagierend bewusstlos Glasgow Coma Scale Eye Opening Response Spontaneous, open with blinking at baseline Opens to verbal command, speech, or shout Opens to pain, not applied to face None Verbal Response

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Oriented

Customization via Module

Entry via:

- Login > Domain Routine > Customise my Modules > click on Master of choice, file builds To edit:
- Scroll to element of choice > click on line of interest > click on **pencil icon** > click on **Edit**
- Change items available
- Use Save, Preview and Discard as appropriate
- To save changes to existing module, click **Save**

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Customization via Table

Entry via:

Login > Domain Routine > Customize my Modules > click on field > choose table* for new version or new module

*For new version or module rename table before upload

For Upload:

Click on field + > Choose File > fill in new Module Name > Submit > Back Button

When count complete, module ready for use

Beispiel Glasgow Coma Scale (GCS)

lasgow Coma Scale				GIGIÉcScGIEsEsSIШグ格拉斯哥智	迷量表		Glasgow C	coma Scale	Header	Title1	
Eye Opening Resp	oonse			EyEyR(RIO)R(R(R(O)開静眼反应				Eye Open	Header	Title2	
	Spontane	ous, open with blinkin	g at baseline	SFSFSFSFSFEESFC自自发的,开	F放的,基线上	有闪烁的	现象	Spontaneo	Button	Radio	GCSEye
	Opens to v	verbal command, spe	ech, or shout	O(O(S'S)O(S(A)O(O)口开启口头部	令、讲话或啊	t话,		Opens to v	Button	Radio	GCSEye
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Verbal Response				V(V(R(R)MR(R(R(Y)口口头回应				Verbal Re	Header	Title2	
	Oriented			01010101G10101Zc01才面向				Oriented	Button	Radio	GCSVerbal
	Confused	conversation, but abl	e to answer questions	CiCiCiCiViCiCiZtCi会谈话混乱。	但能回答问题	Ι.		Confused	Button	Radio	GCSVerbal
	Inappropri	iate responses, words	discernible	In In R(RiO(R)R(NiH)不反应不当,	言语可辨。			Inappropri	Button	Radio	GCSVerbal
	Incomprehensible speech			In In UIDiOIDiDiNiнe不难以理解的	语言			Incompret	Button	Radio	GCSVerbal
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	Obeys cor	mmands for movemen	t	OIOIOIOIGOOIW Re移服从运动排	令			Obeys cor	Button	Radio	GCSMotor
	Purposefu	ul movement to painfu	stimulus	PLPLUM DIM MCLLI痛对疼痛刺激	有目的的运动	b		Purposefu	Button	Radio	GCSMotor
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	Abnormal (spastic) flexion, decorticate posture			AtAtFIFIAtFIFINiHe異异常(痉挛	1性) 屈曲、剱	腹姿势		Abnormal	Button	Radio	GCSMotor
	Extensor ((rigid) response, dece	rebrate posture	E>E>R(R(E)R(R(R(P)伸外展(刚性)反应,去脊柱的姿势				Extensor (Button	Radio	GCSMotor
	None			N(N(ALN)G(N)N(BrH(な无				None	Button	Radio	GCSMotor
GCS Score				GIGIScPIGIPIPcW6εGIGCS分数			GCS Score	GCS Scor	Field	Measurement	

Elemente aufgelistet, Berechnung hinterlegt Im Bericht GCS Score Auflistung der Art von Feldern Verschiedene Sprachen hinterlegt

Anamnese	Untersuch	Prozedere
Allgemein	Alloemeinzustand	
Haut und Nägel	Angemenizostario	
Kopf und Hals	o gut	
Lymphknoten	pathologisch	
Thorax	Details	
Kreislauf	Bowucstsoin	
Abdomen	Dewosstsein	
Neurostatus	🔵 normal	
Bewegungsapparat	 pathologisch 	
	somnolent	
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	Glasgow Coma Scale	
	Eye Opening Response	
	🔵 Spontaneous, open wit	h blinking at baseline
	Opens to verbal comm	and, speech, or shout
	Opens to pain, not app	lied to face
	○ None	
	Verbal Response	
	Oriented	

Diagnoseraster für degenerative Wirbelsäulenerkrankungen

vertebrales Syndrom							
zephales Syndrom							
spondylogenes Syndrom							
	bei/mit						
		Fehlhaltung/Fehlform					
			Lordosierung/ Kopfprotraktion				
			Streckhaltung				
			Kyphosierung				
			Schiefhals/Torticollis				
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Implications of structured data entry

For patients

- Improved and personalized diagnostics
- Access to own data possible
- Structured follow-up data for outcome assessment and quality control

For physicians

- Documentation with discrete, time-stamped data points is available realtime and is paired with reporting,
- Fully addressable and analyzable data for quality control and research
- More face time with patients
- Analysis for individual care, quality control and research

For health professionals

- Structured data entry tailored for disease, as for physicians
- Suggestions for structured diagnosis

For technology

- Data from diverse sources for improved diagnosis, outcome measurement, research
- Option to expand to any other disease

For my institution

 Data to drive process organization and optimization for increased quality and cost savings

Summary

Phenotyping systems for physical characteristics available for research purposes Not operational for clinical routine not given

- Insufficient resolution
- No system for clinical documentation or reporting
- Limited sequential input
- Adaptations resource-intensive
- Limited language editions

Structured data entry with enabled domain implemented adaptations technically feasible for embedding in a given system

What next?

Avatar for medical purposes

Structured data entry

- Health history
- Physical findings

Apparative diagnostics

- Physiologic
- functional

Molecular diagnostics

- Genetics
- Epigenetics
- Other



Routine laboratory

- General
- Specialized

Imaging

- Radiation based
- Ultrasound
- MRI, others
- combined

Response to therapy

- Efficacy, Effectiveness
- Undesired effects