Linking and sharing multi-modal data with Knowledge Graphs

Application to intracranial aneurysms

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Séminaire AISTROSIGHT

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Comprendre les pathologies
cardio-vasculaires, métaboliques et respiratoires pour mieux les **prévenir**

Médecine translationnelle :
Patients ← → Recherche

Lien entre **fonction** biologique et séquences **génétiques** ?

Bio-informatique

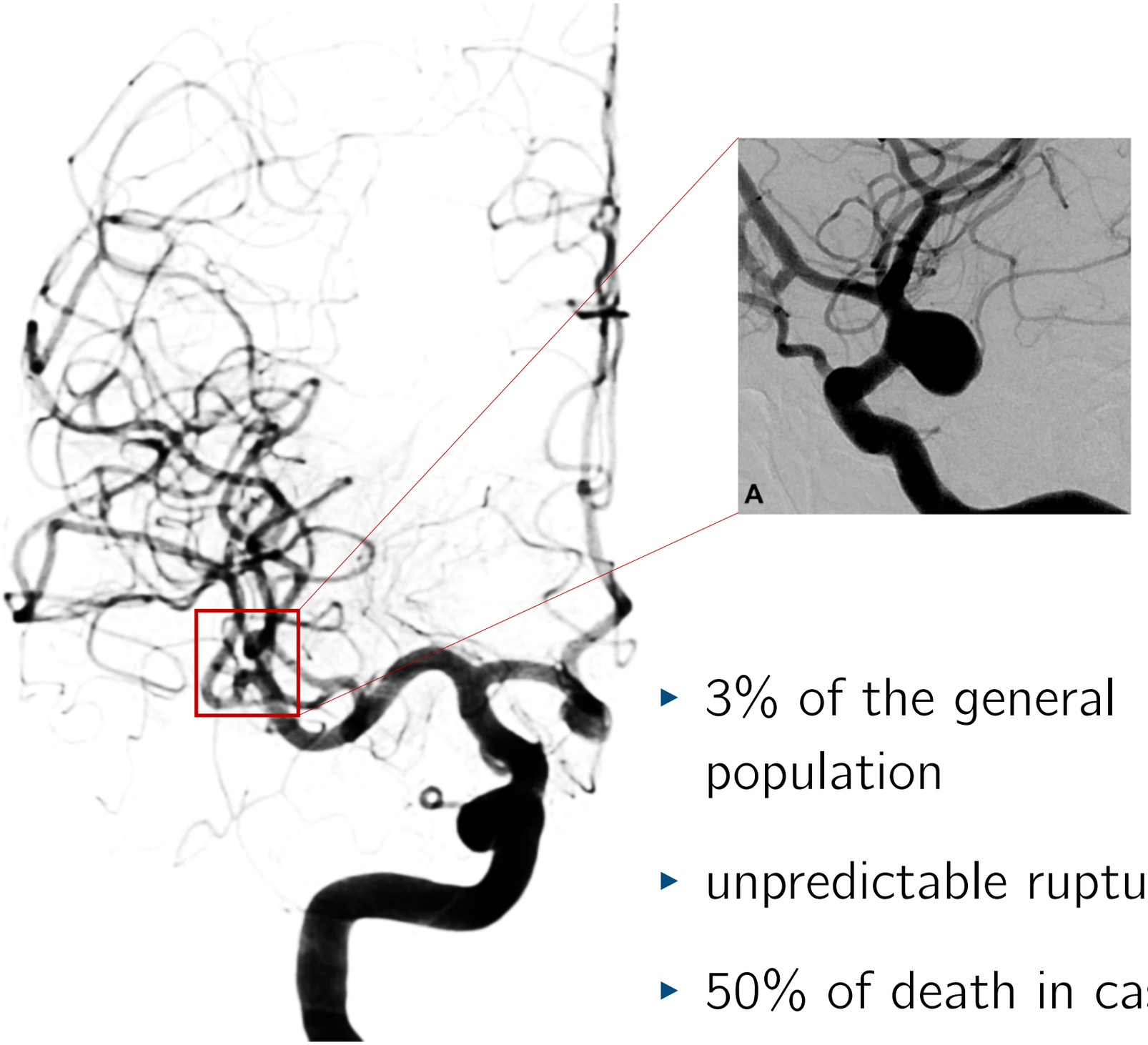
Données de santé en grand nombre

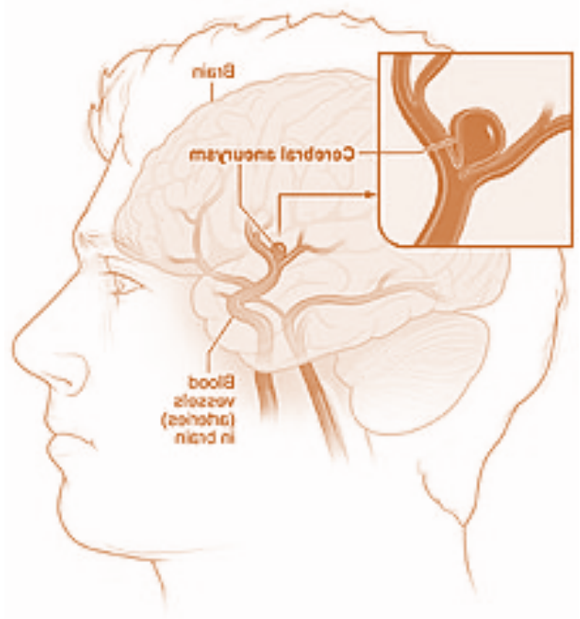
- **calcul haut-débit** : infrastructure distribuées
- **intégration de données** hétérogènes
- **modèles explicatifs / prédictifs (IA)**

données massives → nouvelles connaissances

Intracranial aneurysms

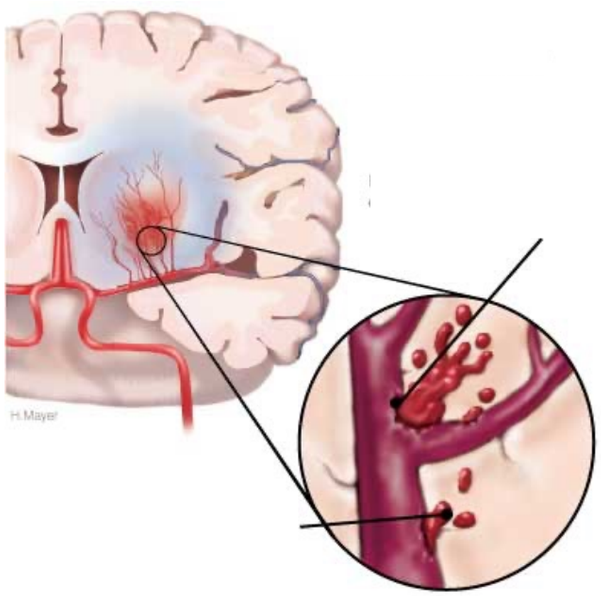
Intracranial aneurysm (ICA)

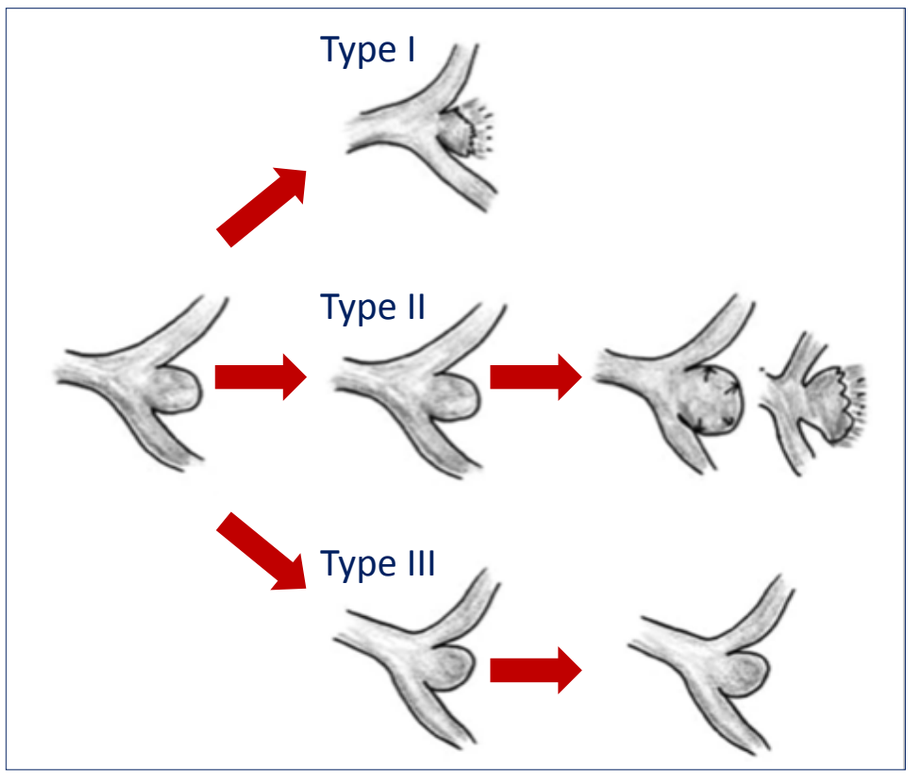




- 3% of the general population
- unpredictable rupture
- 50% of death in case of rupture

ICA rupture / stroke





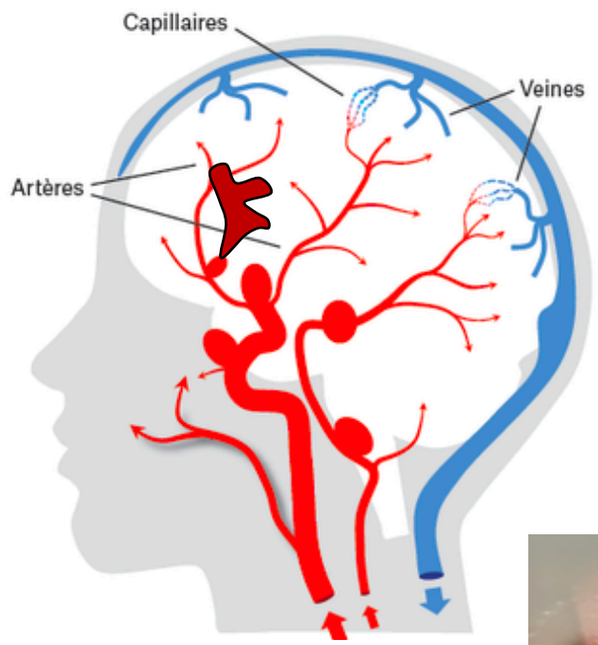
Risk factors

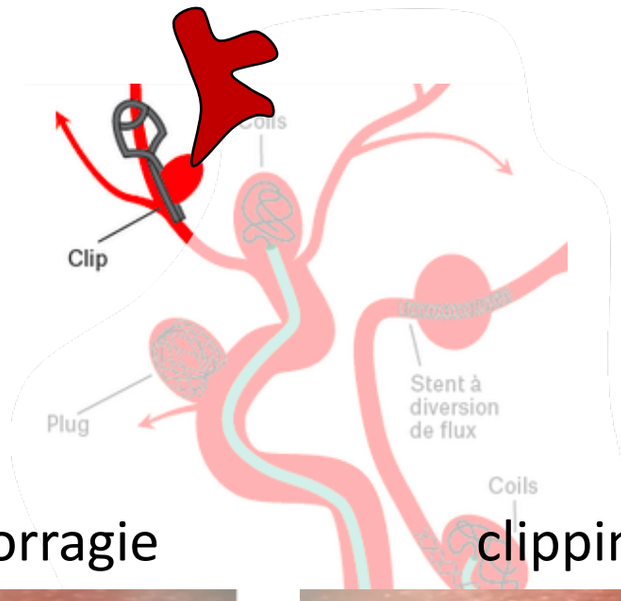
- smocking, alcohol consumption, hypertension, female sex, age
- familial history (RRx4)

In 10% of ICA cases = familial form of AIC with at least 2 first-degree relatives affected

How are they treated ?

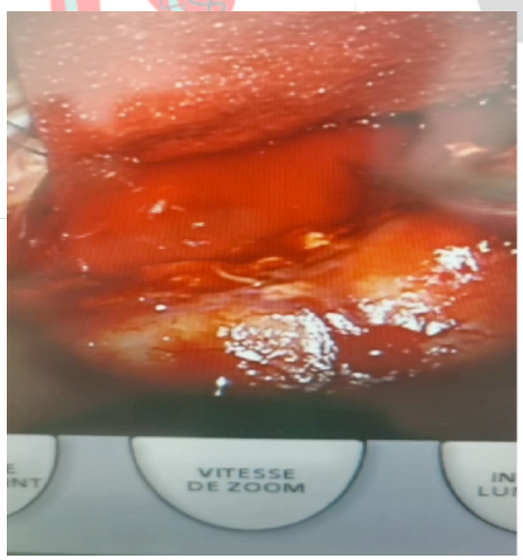
Surgical clipping





Hemorragie

clipping

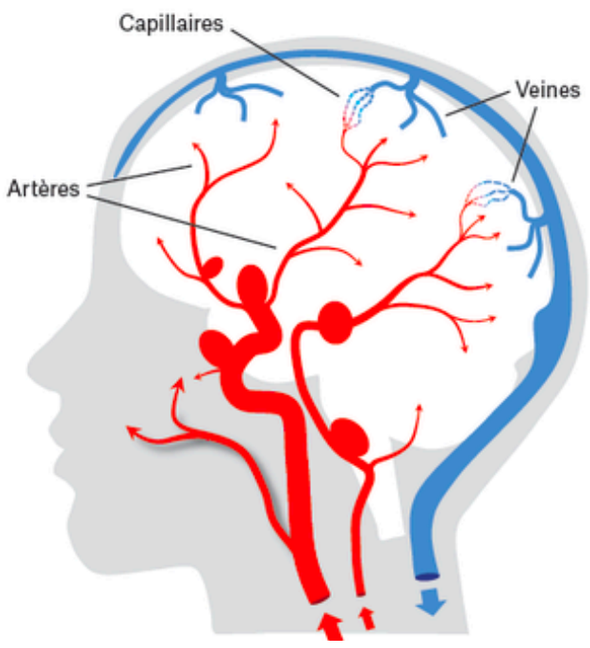


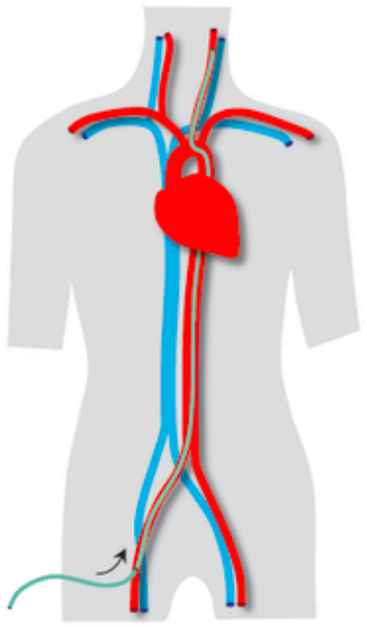
courtesie of Dr Ambrosi – CHU Nantes, Anesthésie-Réanimation

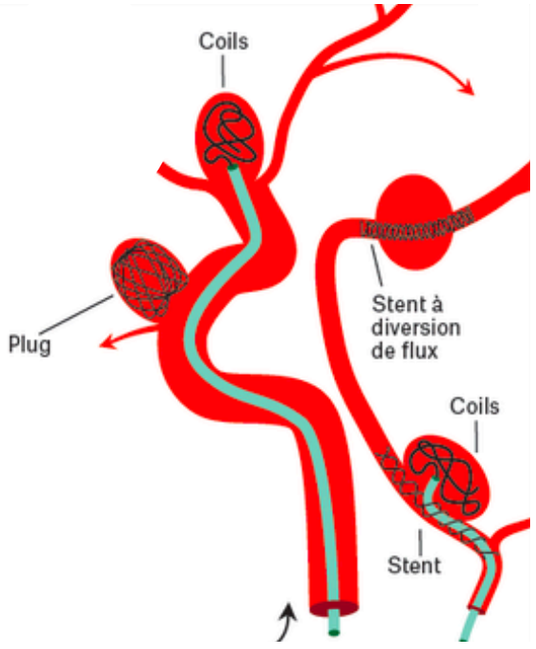
How are they treated ?

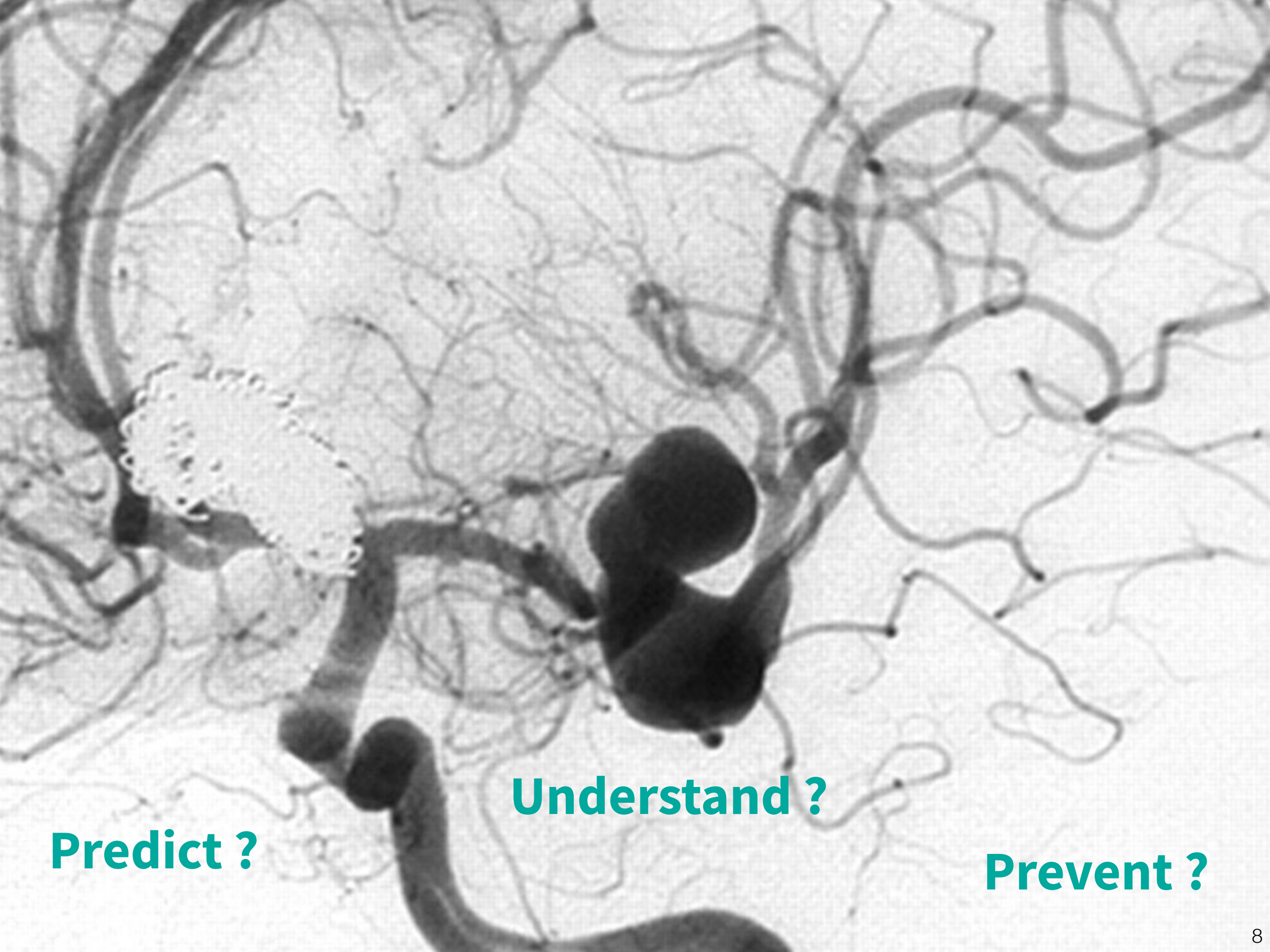
Regular MRI

+ endovascular surgery



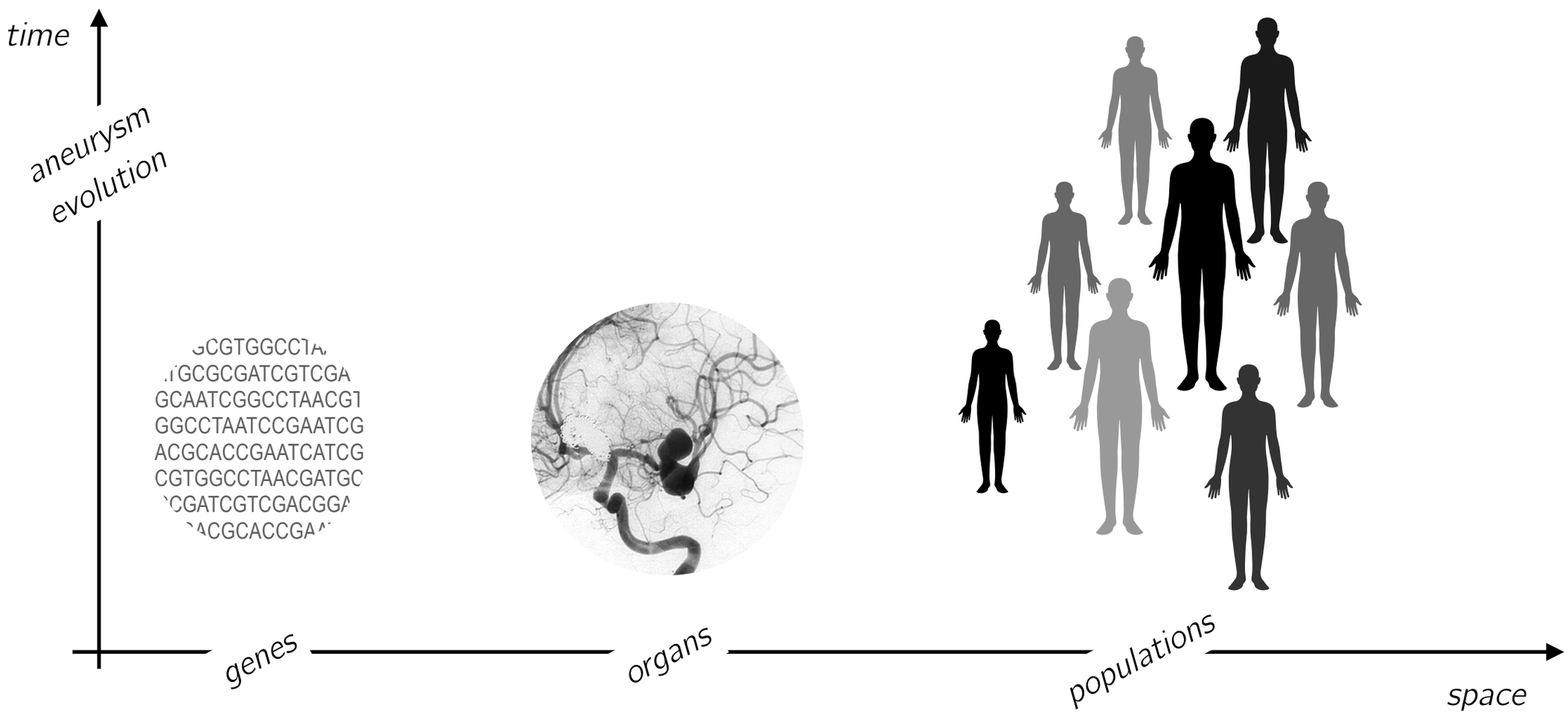





Understand ?
Predict ?
Prevent ?

Multi-factorial disease $\longrightarrow$ multi-scale data

- Intracranial aneurysms: a complex & multifactorial disease
- Inter-disciplicary efforts needed for a better understanding of the pathology
- Specific data produced at very specific scales



Multi-centric study: 29 contributing centers

ICAN, UCAN, weCAN projects

+ recently funded eCAN RHU project

SFNR
SOCIETE FRANCAISE
DE NEURORADIOLOGIE

Fsm
FÉDÉRATION
DES SPÉCIALITÉS MÉDICALES

FRAN

2023

4 000 ICA cases

TOF MRIs

800+ whole genomes

2024 +

> 3 000 patients per year

Multi-modal data analysis

Using deep learning for an automatic detection and classification of the vascular bifurcations along the Circle of Willis

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ARTICLE INFO

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41A10
65D05
65D17

Keywords:
Vascular bifurcations
Circle of Willis
Deep learning

ABSTRACT

Most of the intracranial aneurysms (ICA) occur on a specific portion of the cerebral vascular tree named the Circle of Willis (CoW). More particularly, they mainly arise onto fifteen of the major arterial bifurcations constituting this circular structure. Hence, for an efficient and timely diagnosis it is critical to develop some methods being able to accurately recognize each Bifurcation of Interest (BoI). Indeed, an automatic extraction of the bifurcations presenting the higher risk of developing an ICA would offer the neuroradiologists a quick glance at the most alarming areas. Due to the recent efforts on Artificial Intelligence, Deep Learning turned out to be the best performing technology for many pattern recognition tasks. Moreover, various methods have been particularly designed for medical image analysis purposes. This study intends to assist the neuroradiologists to promptly locate any bifurcation presenting a high risk of ICA occurrence. It can be seen as a Computer Aided Diagnosis scheme, where the Artificial Intelligence facilitates the access to the regions of interest within the MRI. In this work, we propose a method for a fully automatic detection and recognition of the bifurcations of interest forming the Circle of Willis. Several neural networks architectures have been tested, and we thoroughly evaluate the bifurcation recognition rate.

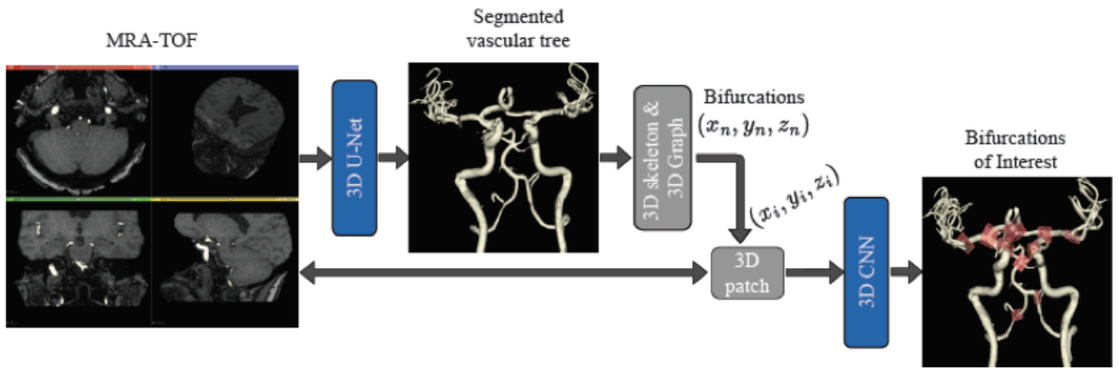


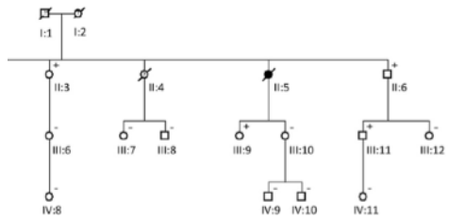
Fig. 1: General flowchart of the bifurcation recognition process.

ARTICLE

Rare Coding Variants in *ANGPTL6* Are Associated with Familial Forms of Intracranial Aneurysm

Romain Bourcier,[1,2] Solena Le Scouarnec,[1] Stéphanie Bonnaud,[1,3] Matilde Karakachoff,[1,3] Emmanuelle Bourcereau,[3] Sandrine Heurtebise-Chrétien,[1] Céline Menguy,[1] Christian Dina,[1,3] Floriane Simonet,[1,3] Alexis Moles,[4] Cédric Lenoble,[2] Pierre Lindenbaum,[1] Stéphanie Chatel,[1,3] Bertrand Isidor,[5] Emmanuelle Génin,[6] Jean-François Deleuze,[7] Jean-Jacques Schott,[1,3] Hervé Le Marec,[1,3] ICAN Study Group, Gervaise Loirand,[1,3,8] Hubert Desal,[1,2,8,*] and Richard Redon[1,3,8,*]

Intracranial aneurysms (IAs) are acquired cerebrovascular abnormalities characterized by localized dilation and wall thinning in intracranial arteries, possibly leading to subarachnoid hemorrhage and severe outcome in case of rupture. Here, we identified one rare nonsense variant (c.1378A>T) in the last exon of *ANGPTL6* (Angiopoietin-Like 6)—which encodes a circulating pro-angiogenic factor mainly secreted from the liver—shared by the four tested affected members of a large pedigree with multiple IA-affected case subjects. We showed a 50% reduction of ANGPTL6 serum concentration in individuals heterozygous for the c.1378A>T allele (p.Lys460Ter) compared to relatives homozygous for the normal allele, probably due to the non-secretion of the truncated protein produced by the c.1378A>T transcripts. Sequencing *ANGPTL6* in a series of 94 additional index case subjects with familial IA identified three other



C

Filtering steps	Remaining variants	
Individuals :	*III-1*	*III-5*
Functional variants	25,674	23,456
MAF < 0,1% in ExAC (NFE)	456	436
Shared by III.1 & III.5	29	
In IBD haplotypes	10	
Shared by all affected	8	
Predicted LOF	1 (*ANGPTL6*)	

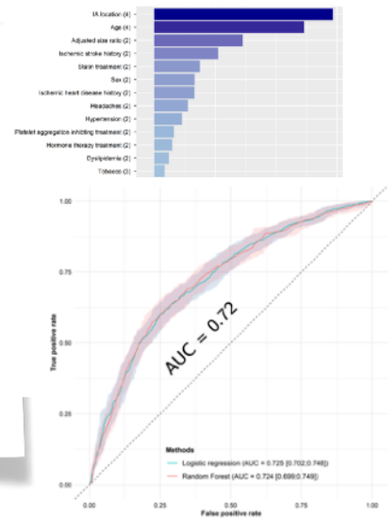
Observational Study > J Neurol Neurosurg Psychiatry. 2021 Feb;92(2):122-128.
doi: 10.1136/jnnp-2020-324371. Epub 2020 Oct 23.

Location of intracranial aneurysms is the main factor associated with rupture in the ICAN population

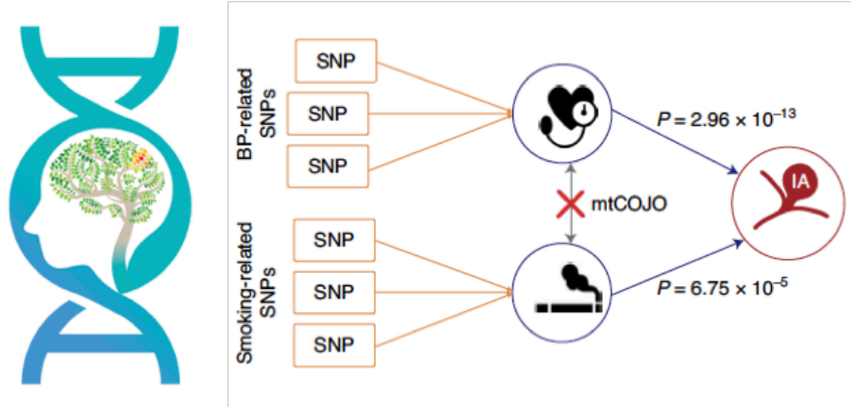
Olivia Rousseau [1], Matilde Karakachoff [1], Alban Gaignard [2], Lise Bellanger [3], Philippe Bijlenga [4], Pacôme Constant Dit Beaufils [1], Vincent L'Allinec [5], Olivier Levrier [6], Pierre Aguettaz [7], Jean-Philippe Desilles [8], Caterina Michelozzi [9], Gaultier Marnat [10], Anne-Clémence Vion [2], Gervaise Loirand [2], Hubert Desal [11], Richard Redon [2], Pierre-Antoine Gourraud [1], Romain Bourcier [12]; ICAN Investigators

Collaborators, Affiliations + expand

PMID: 33097563 DOI: 10.1136/jnnp-2020-324371



Genome-wide association study of intracranial aneurysms identifies 17 risk loci and genetic overlap with clinical risk factors

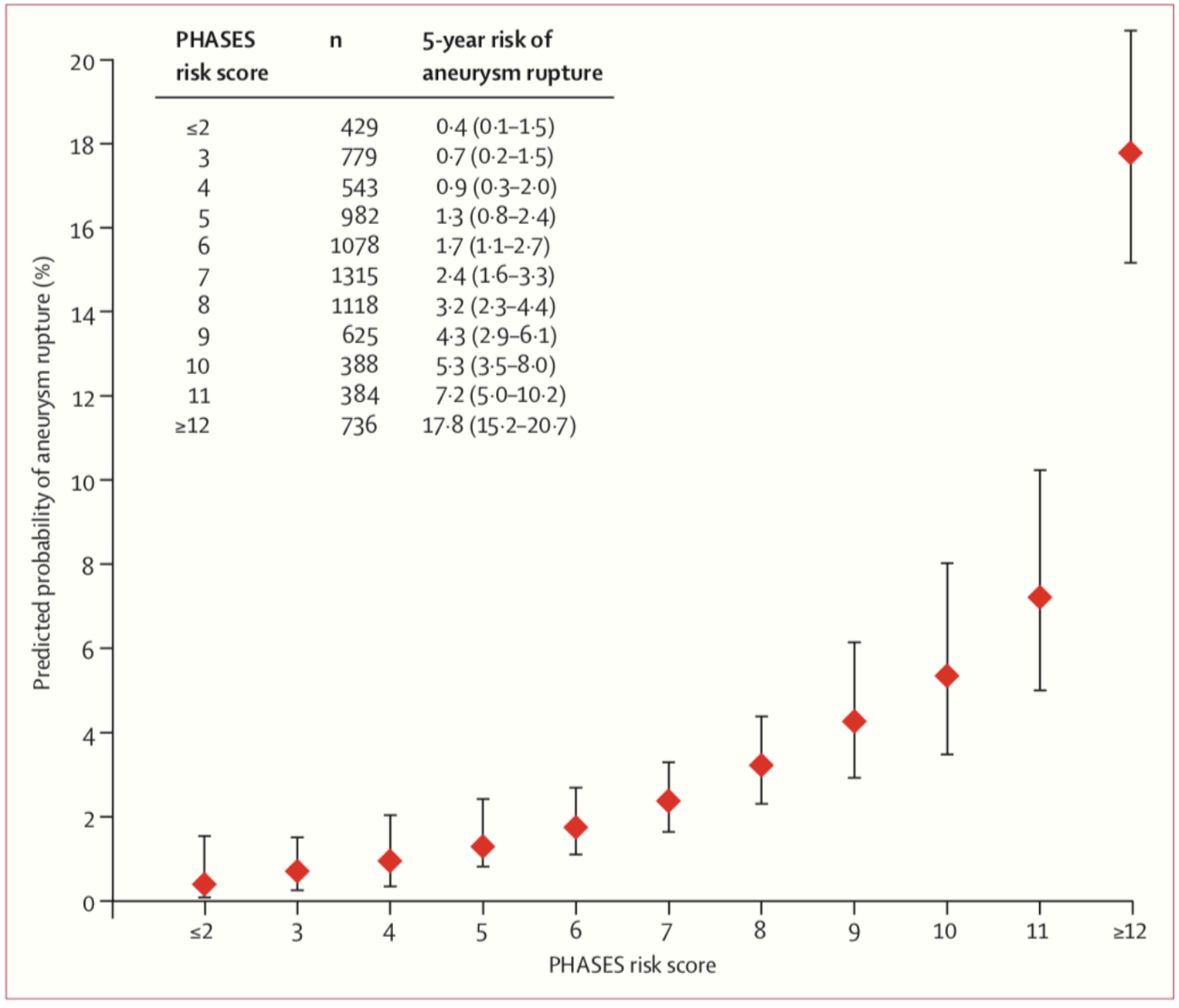


Olivia Rousseau

Is it possible to **predict** IA **rupture** from **clinical data** ?

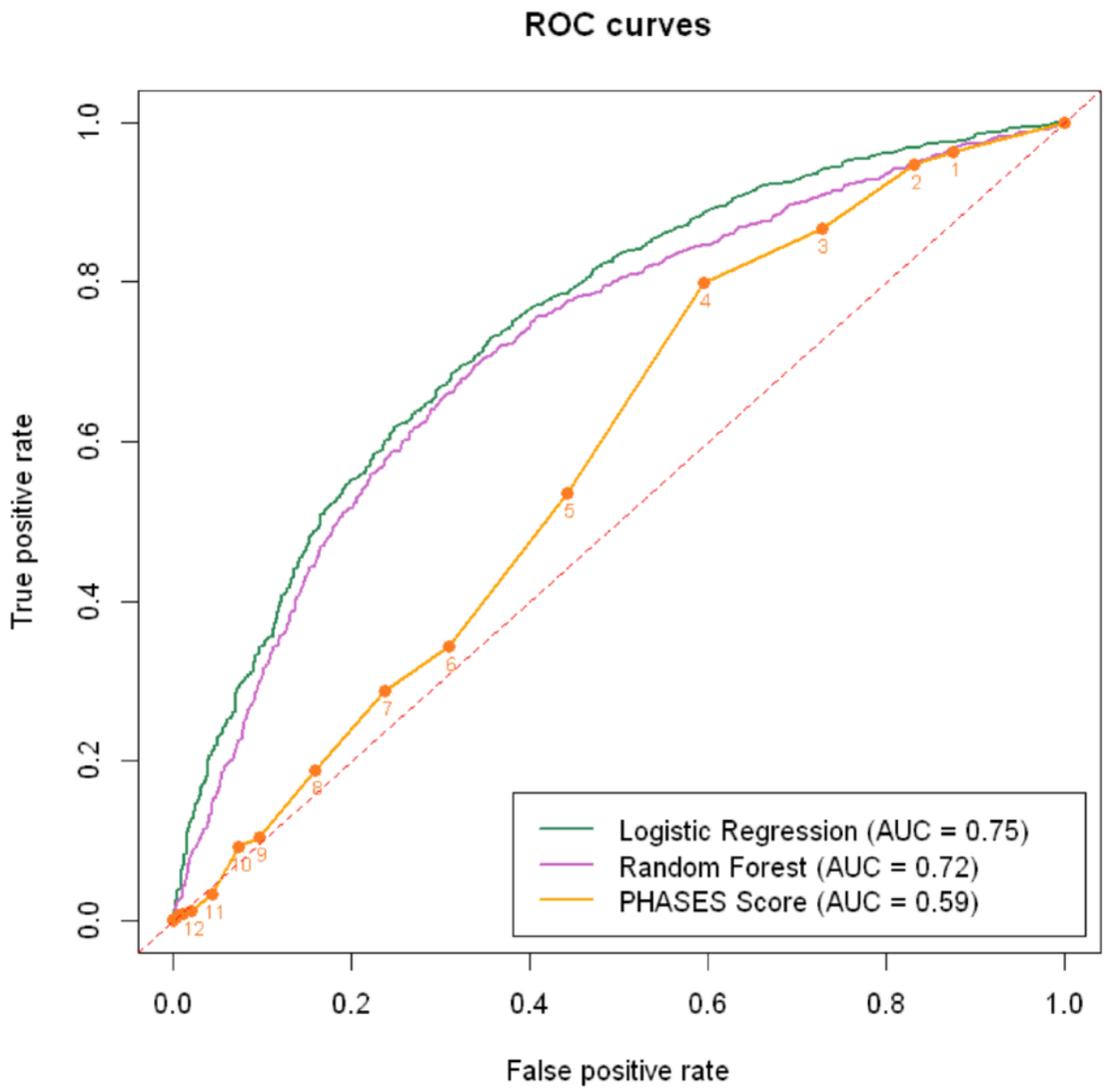
PHASES score

PHASES aneurysm risk score	Points
(P) Population	
North American, European (other than Finnish)	0
Japanese	3
Finnish	5
(H) Hypertension	
No	0
Yes	1
(A) Age	
<70 years	0
≥70 years	1
(S) Size of aneurysm	
<7·0 mm	0
7·0–9·9 mm	3
10·0–19·9 mm	6
≥20 mm	10
(E) Earlier SAH from another aneurysm	
No	0
Yes	1
(S) Site of aneurysm	
ICA	0
MCA	2
ACA/Pcom/posterior	4



Greving et. al. 2014 : Development of the PHASES score for prediction of risk of rupture of intracranial aneurysms: a pooled analysis of six prospective cohort studies.

Applying PHASES score to ICAN clincal data



- Unfortunately, poor prediction accuracy for PHASES (0.59)
- Better prediction accuracy with state-of-the-art Machine Learning / Statistical methods + other variables
- **Predictions are still failing in around 25% of cases …**

integrating imaging or genomic markers ?

Rousseau O, Karakachoff M, Gaignard A, et al Location of intracranial aneurysms is the main factor associated with rupture in the ICAN population Journal of Neurology, Neurosurgery & Psychiatry Published Online First: 23 October 2020. doi: 10.1136/jnnp-2020-324371

Intracranial aneurysms, current challenges

Complex and **multi-scale** biological phenomenons involved :

- Impact of life habits ? Image-based evaluation ?
- Any genetic variant specific to image-based phenotypes ?
- Mirror aneurysms patient sub-grouping ? (Vincent Lallinec)

Needs for **jointly** exploiting genomic data, MR imaging and clinical observations …

… to better **understand** and **predict** evolution schemes.

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39458_2007      39458_2009      39458_2012      39458_3003      39458_39461     39458_39462     39458_39465     39458_39466
39458_CD12099   39458_CD12100   39458_CD12252   39458_CD12350   39458_CD12380   39458_CD12406   39458_CD12466   39458_CD12480
39458_CD12501   39458_CD12586   39458_CD12587   39458_CD12588   39458_CD12704   39458_CD13016   39458_CD13018   39458_CD13178
39458_CD13363
1       752566  rs3094315       A       G       .       .       PR      GT      ./.     ./.     ./.     ./.     ./.     ./.
./.     ./.     ./.     ./.     ./.     0/1     0/0     0/1     0/1     0/1     0/0     0/1     0/1     0/1     0/1     0/1
0/1     0/0     0/0     0/1     0/1     1/1
1       754105  rs12184325      C       .       .       .       PR      GT      ./.     ./.     ./.     ./.     ./.     ./.
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0/0     0/0     0/0     0/0     0/0     0/0
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0/1     0/0     0/0     0/1     0/1     1/1
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0/0     0/0     0/0     0/0     0/0     0/0
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0/1     0/0     0/0     0/0     0/1     0/0
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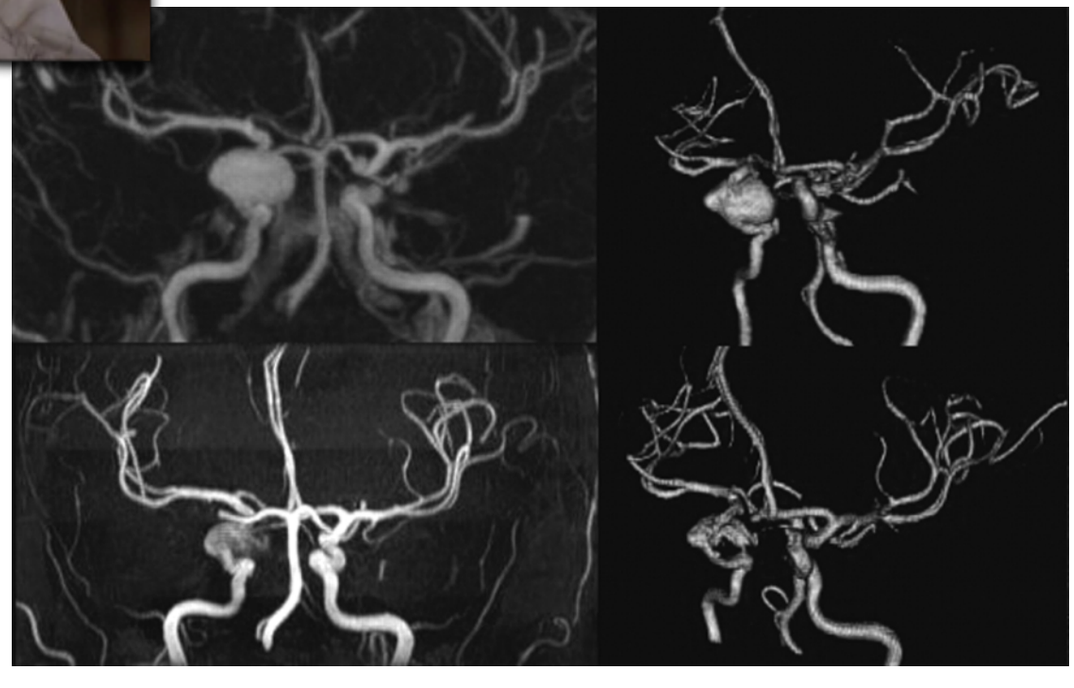
nts atteint	circonstances di	consommation c	quantite alcool p	statut phenotyp	nb d anevrismes	date du 1er diag	AIC 1 Grand axe	AIC 1 Localisatic	AIC 2 Grand axe	AIC
	Fortuite	Sevrage > 3ans	96.6	certain	1.0	1/22/16	7.0	5		
	Dépistage familial	Tabagisme actif	277.2	certain	1.0	3/3/16	4.0	16		
	Fortuite	Sevrage < 3 ans	108.2	certain	3.0	12/29/15	6.0	4	14.0	1
	Rupture AIC	Jamais fumé	0.0	certain	2.0	4/3/09	7.0	M.C.A.Gche	5.0	M.C.A.D
	Rupture AIC	Sevrage > 3ans		certain	4.0	1/1/97		18		17
	Fortuite	Jamais fumé	25.2	certain	1.0	1/5/16	5.0	21		
	Fortuite	Sevrage < 3 ans	50.4	certain	3.0	9/5/15	5.0	18	8.9	118
	Rupture AIC	Jamais fumé		certain	1.0	5/1/05	6.0	118		
	Rupture AIC	Jamais fumé		certain	1.0	10/16/10	9.0	107		
	Fortuite	Tabagisme actif		certain	1.0		7.0	105		
	Rupture AIC	Sevrage > 3ans	39.8	certain	1.0	1/5/10	2.0	10		
	Rupture AIC	Jamais fumé		certain	1.0		4.0	10		
	Fortuite	Jamais fumé	16.2	certain	1.0		8.0	10		
	Fortuite	Sevrage > 3ans		certain	2.0		8.0	18	5.0	18
	Fortuite	Jamais fumé		certain	1.0	8/13/16	26.0	104		
	Dépistage familial	Jamais fumé		certain	1.0	1/1/98	8.0	118		
	Fortuite	Tabagisme actif		certain	1.0		8.0	20		
	Rupture AIC	Tabagisme actif	25.2	certain	4.0	3/3/18	7.0	17	4.0	104
	Rupture AIC	Tabagisme actif	396.0	certain	1.0		5.0	132		
	Rupture AIC	Jamais fumé		certain	3.0	1/1/10	3.0	10	4.0	10
	Rupture AIC	Tabagisme actif		certain	2.0	9/23/05	12.0	10	3.0	100
	Dépistage familial	Sevrage < 3 ans	378.0	certain	1.0	1/1/16	2.0	18		
	Rupture AIC	Jamais fumé		certain	1.0	2/5/12	25.0	6		
	Rupture AIC	Jamais fumé		certain	1.0	1/1/03	6.0	6		
	Rupture AIC	Tabagisme actif	84.0	certain	3.0	10/1/16	4.0	10	2.0	118
	Rupture AIC	Jamais fumé		certain	2.0	3/15/16	5.0	118	1.5	18
	Dépistage familial	Sevrage > 3ans		certain	1.0		2.0	18		
	Rupture AIC	Tabagisme actif	0.0	certain	3.0	6/14/16	2.0	18	5.0	21
		Sevrage > 3ans	12.6	certain	3.0	1/1/14	8.0	10	2.0	18
	Rupture AIC	Sevrage > 3ans	25.2	certain	1.0	6/21/16	2.0	10		
	Fortuite	Jamais fumé		certain	1.0	12/5/16				
	Rupture AIC	Sevrage < 3 ans	170.4	certain	2.0	3/24/15	2.5	10	3.0	12
	Fortuite	Tabagisme actif	12.6	certain	1.0	1/1/98	8.0	10		
	Fortuite	Tabagisme actif	0.0	certain	2.0	7/15/15	11.0	118	5.5	18
	Fortuite	Jamais fumé		certain	1.0	12/18/14	7.0	10		
	Rupture AIC	Jamais fumé	0.0	certain	4.0			18	4.0	105
		Jamais fumé	0.0	certain	1.0					
		Jamais fumé	0.0	certain	2.0		3.5	12	2.5	18
	Fortuite	Jamais fumé		certain	1.0	7/23/12	5.0	132		
	Rupture AIC	Jamais fumé	0.0	certain	2.0	4/24/15	5.5	105	13.0	5
	Fortuite	Tabagisme actif	42.0	certain	1.0	1/1/15	7.0	105		
	Dépistage familial	Sevrage < 3 ans	21.0	certain	1.0	6/23/16	9.0	109		
	Fortuite	Tabagisme actif	11.76	certain	2.0	6/5/16	2.8	5	3.0	105
	Rupture AIC	Sevrage < 3 ans	35.28	certain	1.0	9/4/18	5.9	111		
	Fortuite	Sevrage > 3ans	0.0	certain	1.0	4/1/18	8.5	121		
	Rupture AIC	Jamais fumé	11.76	certain	1.0	2/13/14	8.5	10		
	Rupture AIC	Sevrage > 3ans	35.28	certain	1.0	9/5/15	6.7	10		
	Fortuite	Jamais fumé	11.76	certain	1.0	2/22/16	6.0	111		
	Dépistage familial	Tabagisme actif	11.76	certain	1.0	4/1/15	9.0	107		
	Fortuite	Sevrage > 3ans	260.20	certain	2.0	1/1/14	4.4	17	5.2	18



How to jointly analyse these diverse datasets ?

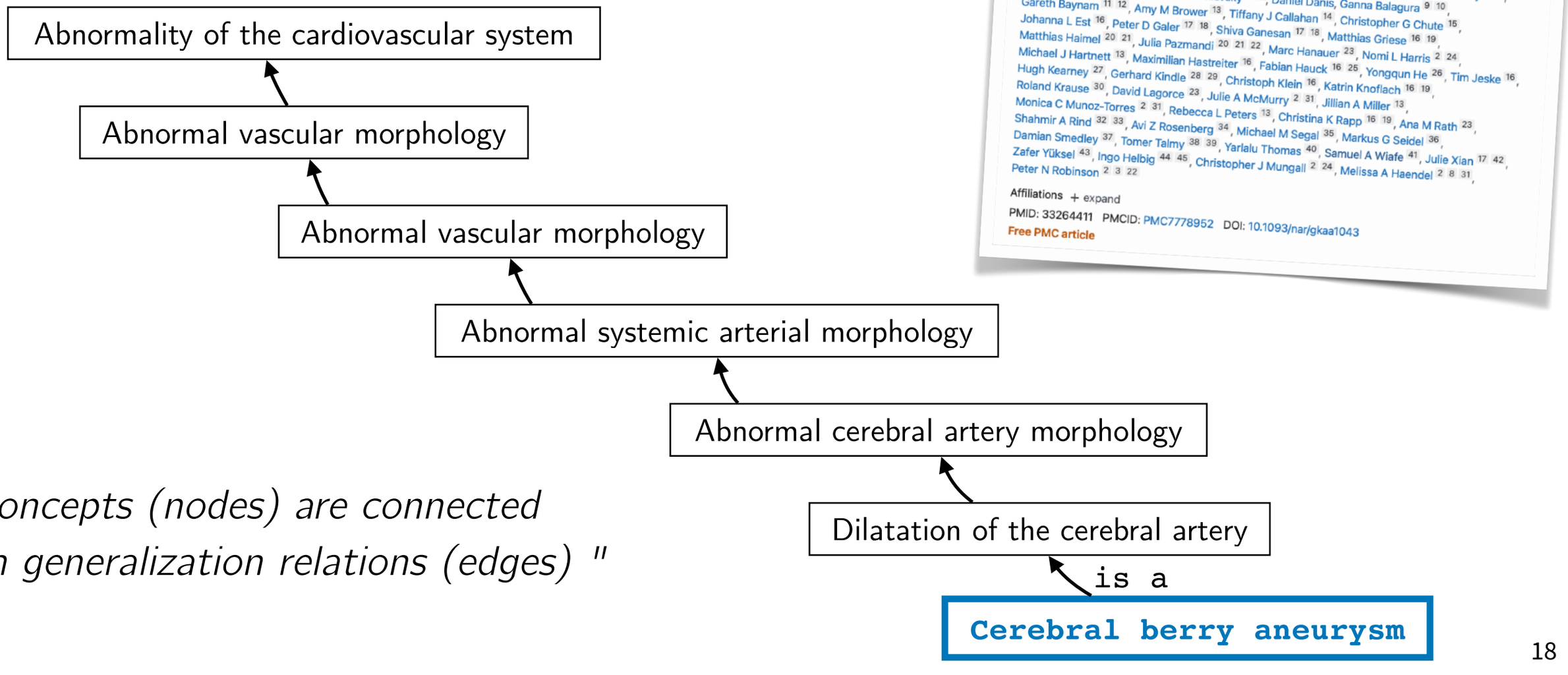
❶ **what** is observed with data ?

Computational ontology

« a formal specification of a shared conceptualization » (Borst, 1997)

→ 1,049 life science ontologies registered in BioPortal (2023)

Human Phenotype Ontology



" Concepts (nodes) are connected with generalization relations (edges) "

> Nucleic Acids Res. 2021 Jan 8;49(D1):D1207-D1217. doi: 10.1093/nar/gkaa1043.

The Human Phenotype Ontology in 2021

Sebastian Köhler [1 2], Michael Gargano [2 3], Nicolas Matentzoglu [2 4 5], Leigh C Carmody [2 3], David Lewis-Smith [6 7], Nicole A Vasilevsky [2 8], Daniel Danis, Ganna Balagura [9 10], Gareth Baynam [11 12], Amy M Brower [13], Tiffany J Callahan [14], Christopher G Chute [15], Johanna L Est [16], Peter D Galer [17 18], Shiva Ganesan [17 18], Matthias Griese [16 19], Matthias Haimel [20 21], Julia Pazmandi [20 21 22], Marc Hanauer [23], Nomi L Harris [2 24], Michael J Hartnett [13], Maximilian Hastreiter [16], Fabian Hauck [16 25], Yongqun He [26], Tim Jeske [16], Hugh Kearney [27], Gerhard Kindle [28 29], Christoph Klein [16], Katrin Knoflach [16 19], Roland Krause [30], David Lagorce [23], Julie A McMurry [2 31], Jillian A Miller [13], Monica C Munoz-Torres [2 31], Rebecca L Peters [13], Christina K Rapp [16 19], Ana M Rath [23], Shahmir A Rind [32 33], Avi Z Rosenberg [34], Michael M Segal [35], Markus G Seidel [36], Damian Smedley [37], Tomer Talmy [38 39], Yarlalu Thomas [40], Samuel A Wiafe [41], Julie Xian [17 42], Zafer Yüksel [43], Ingo Helbig [44 45], Christopher J Mungall [2 24], Melissa A Haendel [2 8 31], Peter N Robinson [2 3 22]

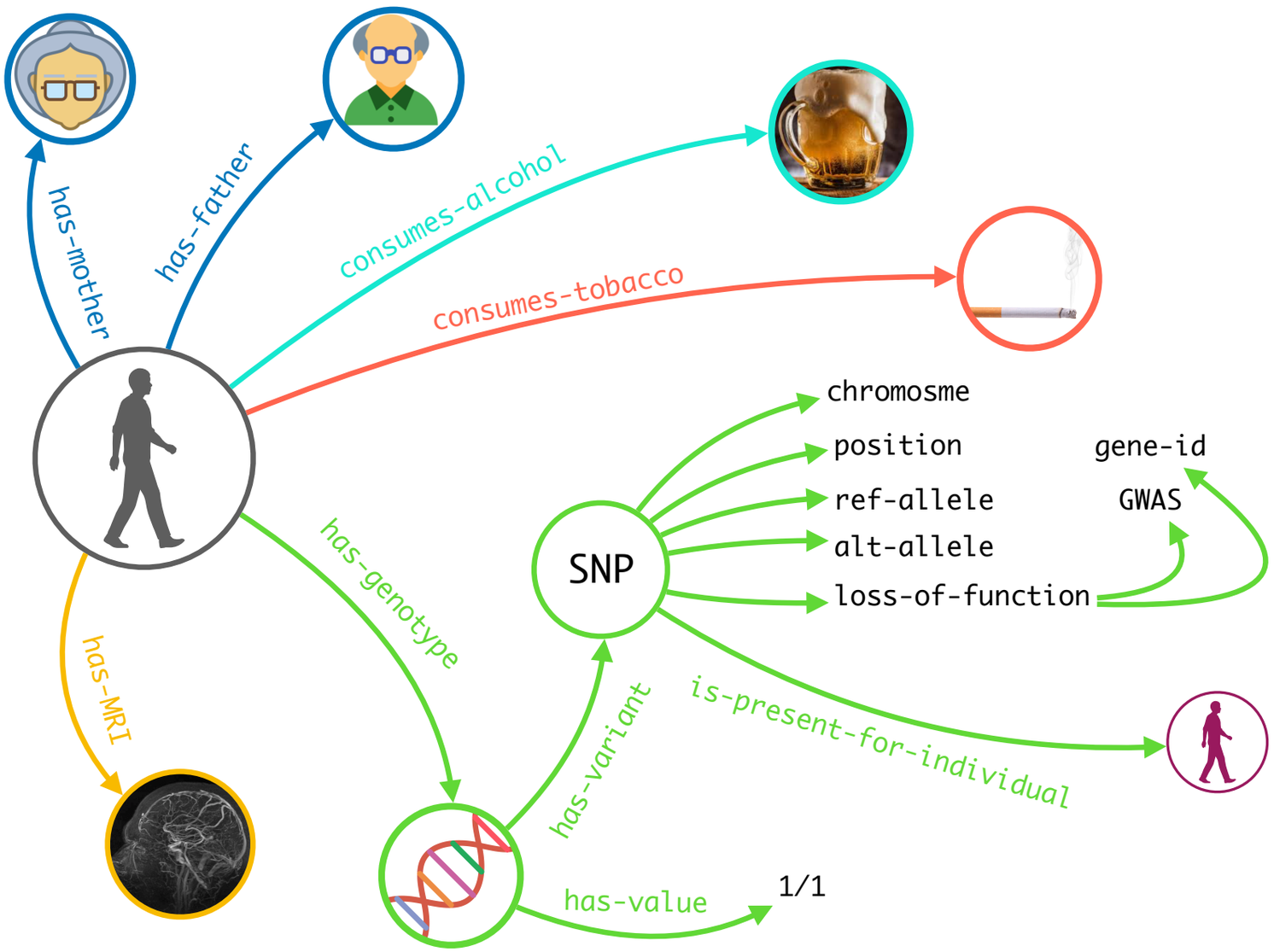
Affiliations + expand

PMID: 33264411 PMCID: PMC7778952 DOI: 10.1093/nar/gkaa1043

Free PMC article

❷ how to **link** data from multiple scales ?

Knowledge graphs: « a collection of interlinked descriptions of things (real-word objects, abstract concepts, events, etc.) »



= **database**: information storage / extraction

= **graph**: network analysis

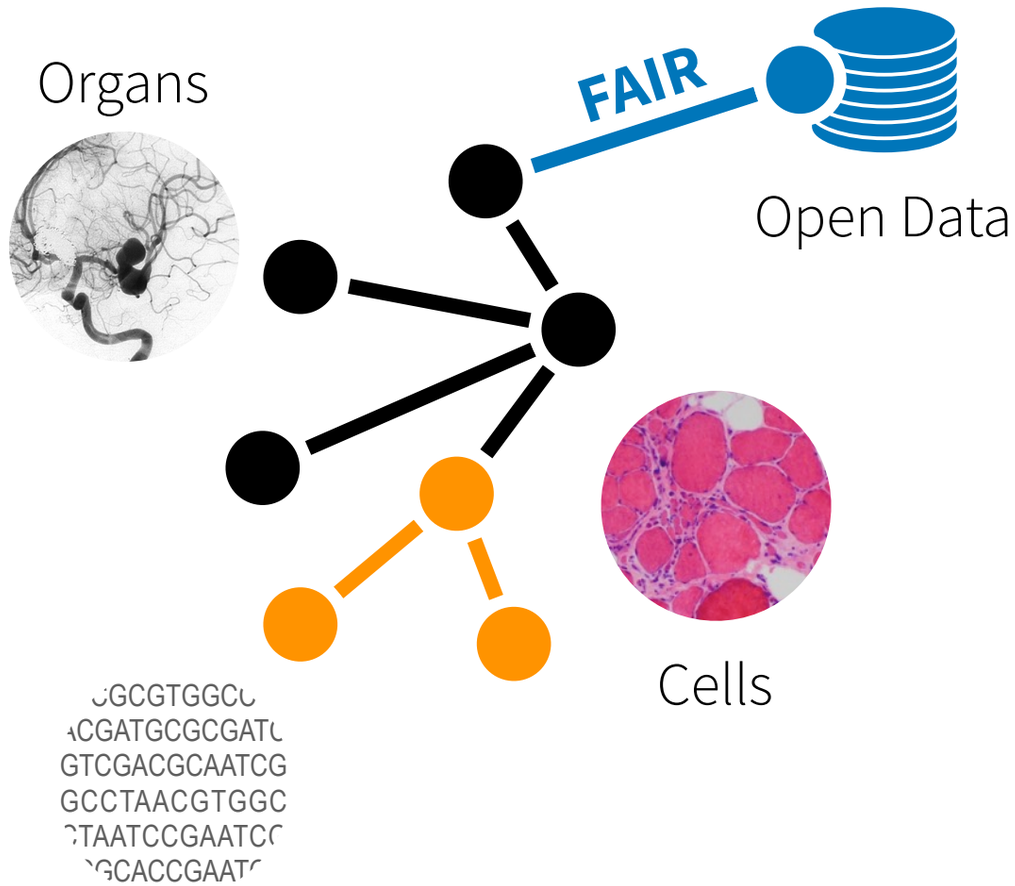
= **knowledge base** with formal semantics :

+ logical facts

+ logical inferences

Bridging imaging-omics-clinical data

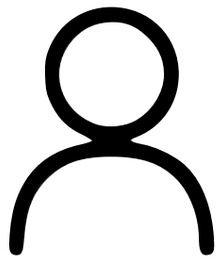
INEX-MED



Multi-scale « Knowledge Graph »

145 Million facts

Clinical observations



- Aim : representing key characteristics of ICA patients

Person
sub_class_of
...
has_center
has_clinical_validation
...
has_gender
has_size
has_weight
has_bmi
has_number_aneurysms
...
is_sporadic
has_phenotype
rupture
Subject
has_tobacco
Tobacco consumption
duration_in_years
packs_a_day
packs_a_year
status
...
has_history_carotid_artery_stenosis
has_history_ischemic_artery_disease
has_history_ischemic_stroke
has_history_peripheral_artery_disease
has_alcohol
Alcohol consumption
absorbed_quantity_g
...
has_aneurysm
has_hypertension
has_diabetes
has_dyslipidemia
Aneurysm
has_size
has_location
has_been_treated
has_ruptured
...

Genomic markers

- Aim : representing associated genetic variations by **extending reference ontologies** (Sequence Ontology, VARIO)

so:0000694
(SNP)

is_a

Variant

has_chromosome
has_position
has_ref_allele
has_alt_allele
has_quality_score
has_gene_ensembl_id
has_gene_name
…

reported_in_GWAS

GWAS

has_trait
has_pvalue
has_pubmed_id
…

Person

sub_class_of

Subject

has_biological_sample

Sample

is_heterozygotous
is_homozygotous_ref
is_heterozygotous_alt

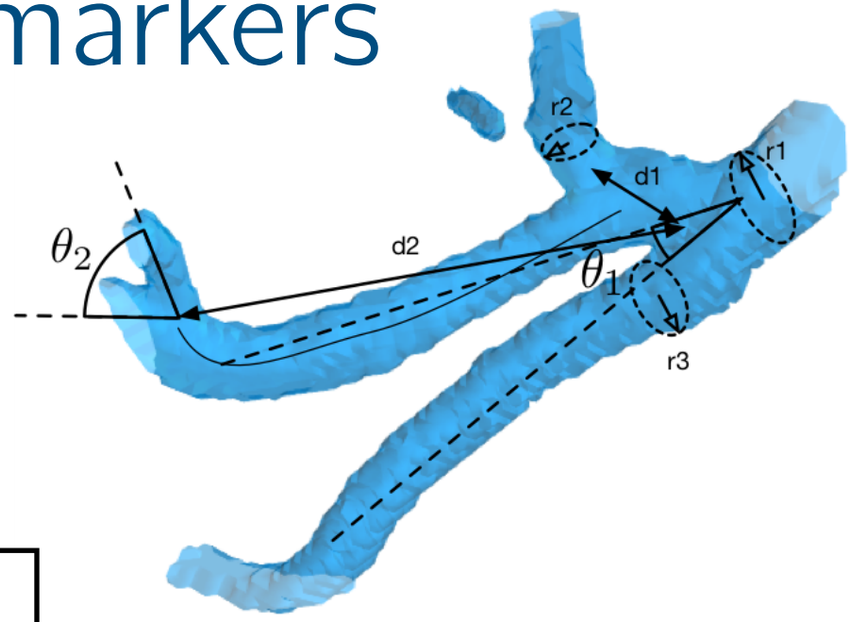
has_loss_of_function

Loss_Of_Function

sub_class_of

vario:0043

Imaging markers



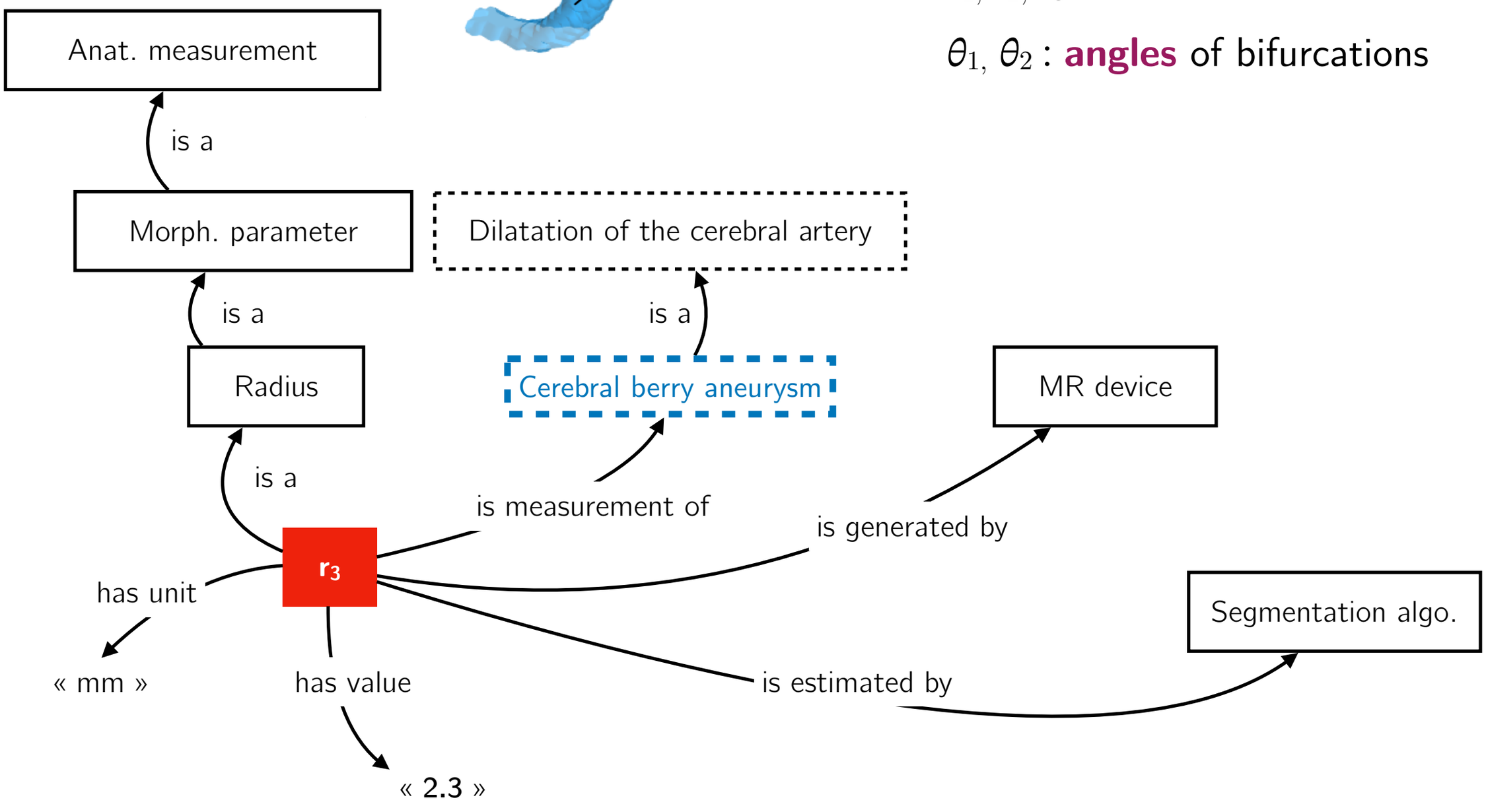
Anass Nouri

Florent Autrusseau

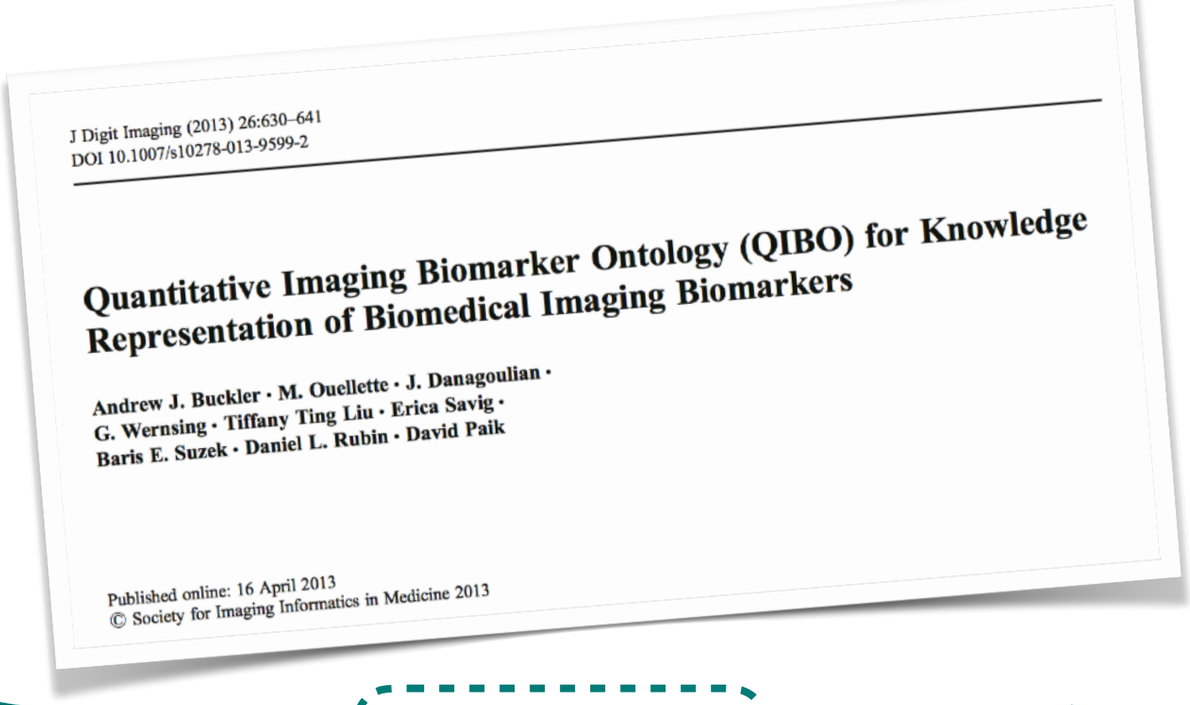
d_1, d_2 : **distances** between **bifurcations**

r_1, r_2, r_3 : **radius** of vessels

θ_1, θ_2 : **angles** of bifurcations



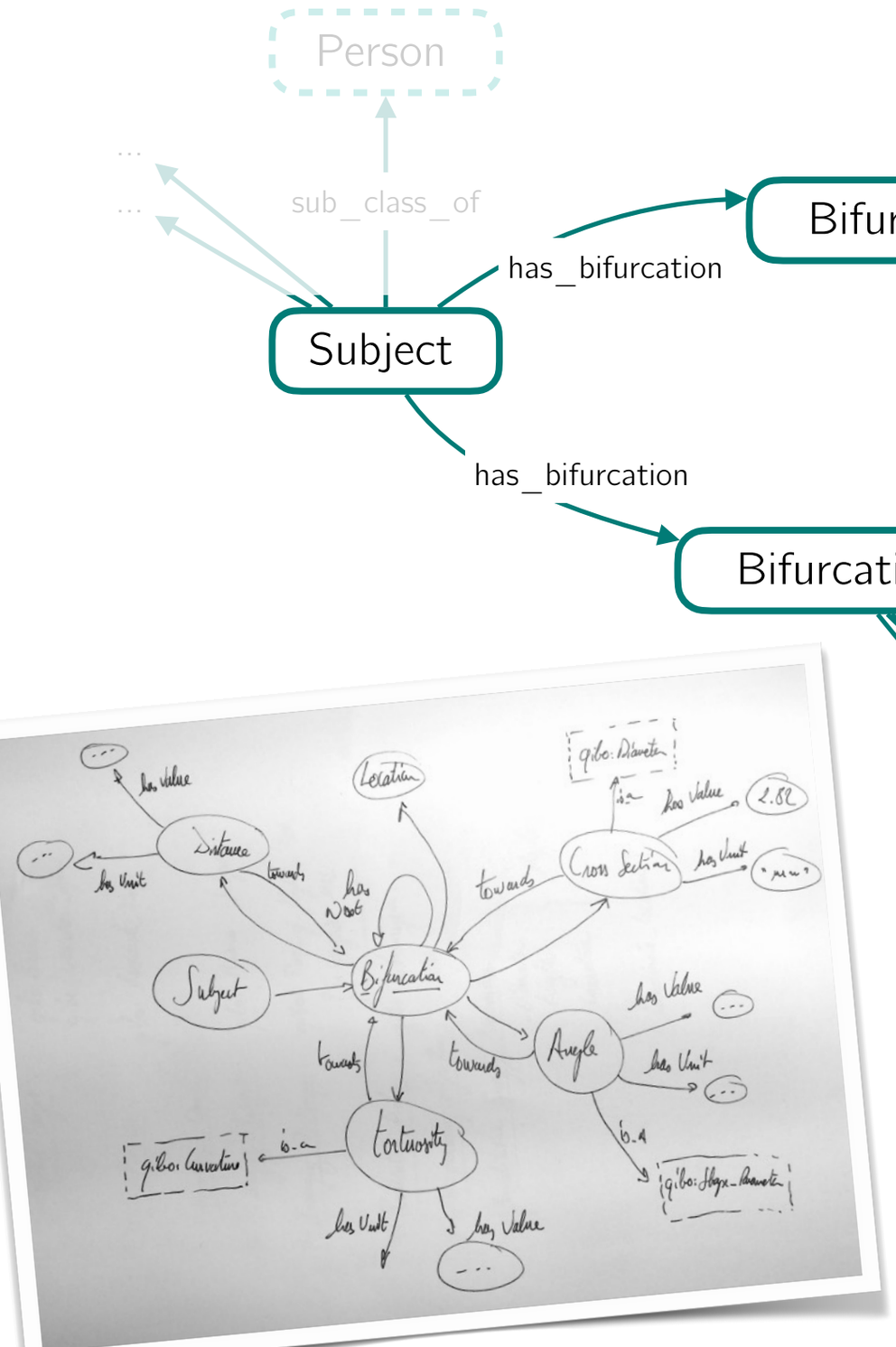
Imaging markers


J Digit Imaging (2013) 26:630–641
DOI 10.1007/s10278-013-9599-2

Quantitative Imaging Biomarker Ontology (QIBO) for Knowledge Representation of Biomedical Imaging Biomarkers

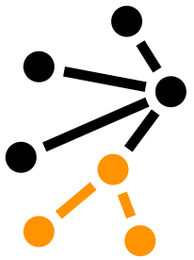
Andrew J. Buckler · M. Ouellette · J. Danagoulian · G. Wernsing · Tiffany Ting Liu · Erica Savig · Baris E. Suzek · Daniel L. Rubin · David Paik

Published online: 16 April 2013
© Society for Imaging Informatics in Medicine 2013



Searching multi-scale data ?

Queries with graph patterns



? *"Give all patients with a mutation in ANGPTL6, in the case of a rupture, give me also the age and the location of the ruptured aneurysm."*

```
SELECT * WHERE {
  # Search variants
  ?subject ican:has_dna_sample ?sample .
  ?sample ican:is_heterozygotous ?variant .
  ?variant ican:has_chromosome "19" ;
           ican:has_position ?position ;
           ican:has_alt_allele ?alt_allele ;
           ican:has_ref_allele ?ref_allele .
  FILTER (?position > 10092337 && ?position < 10106407)
  # dbSNP identifier
  OPTIONAL { ?variant ican:has_dbsnp_id ?dbSNPid . }
  # First aneurysm rupture: age and localisation
  OPTIONAL { ?subject ican:age_first_rupture ?ageRupt . }
  OPTIONAL { ?subject ican:loc_first_rupture ?locRupt . }
}
```

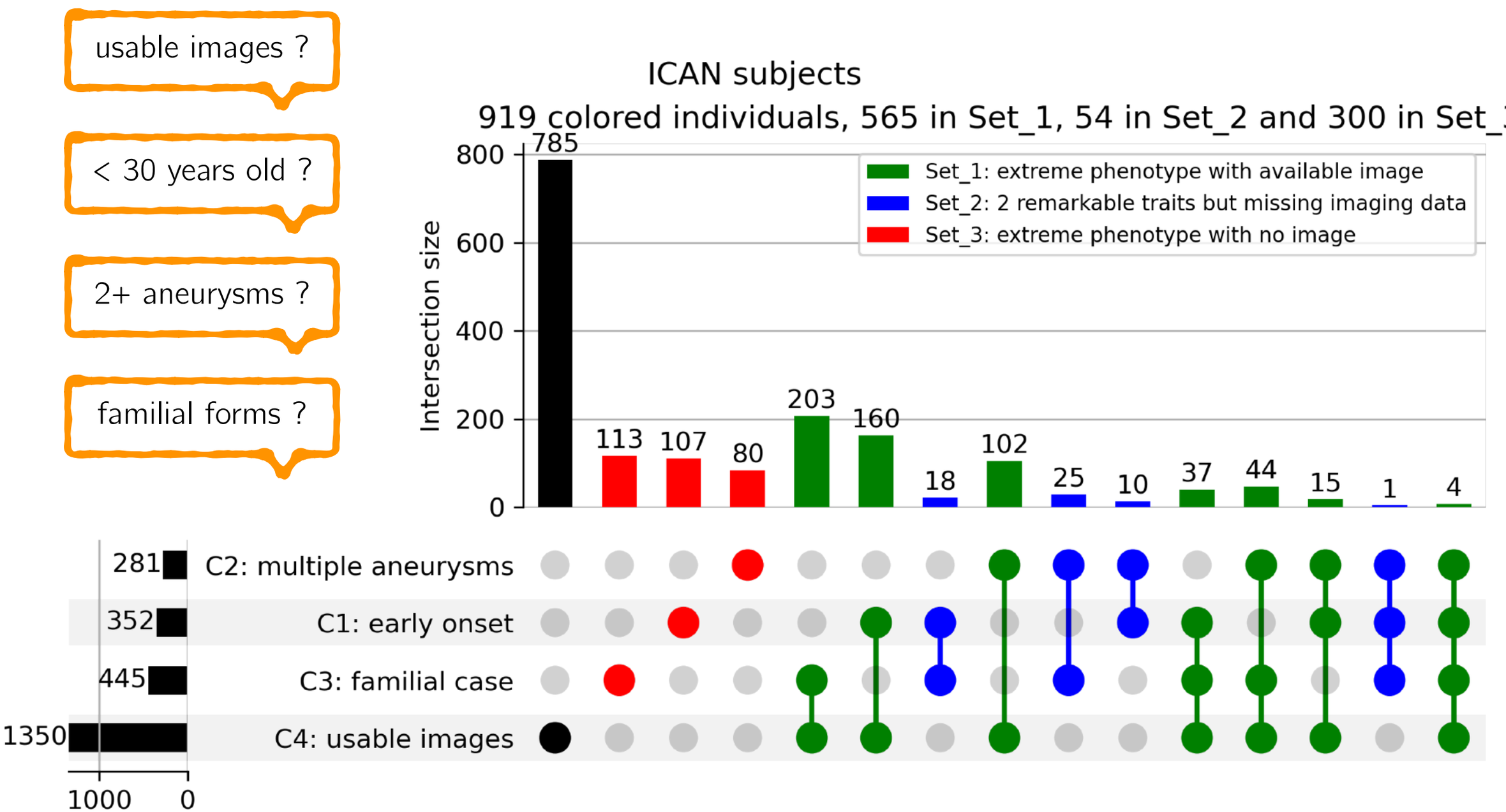
Select "extreme phenotypes" for genomic studies

usable images ?

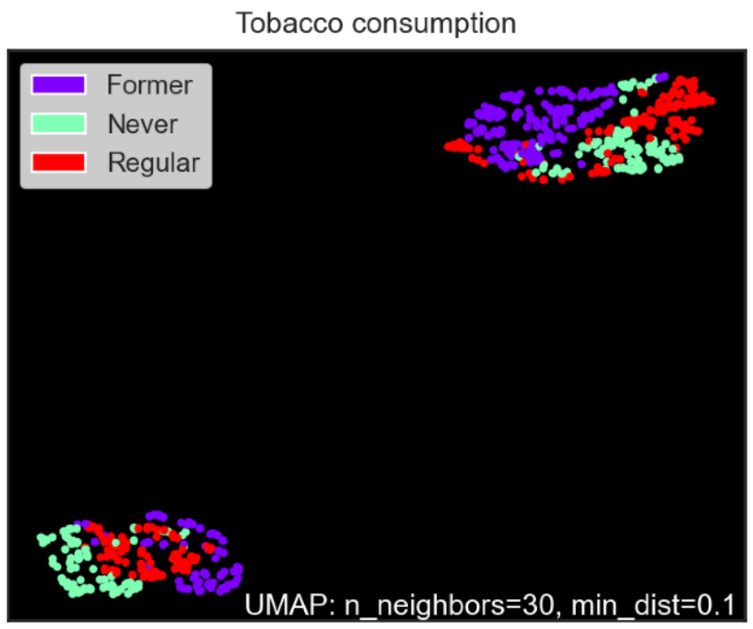
< 30 years old ?

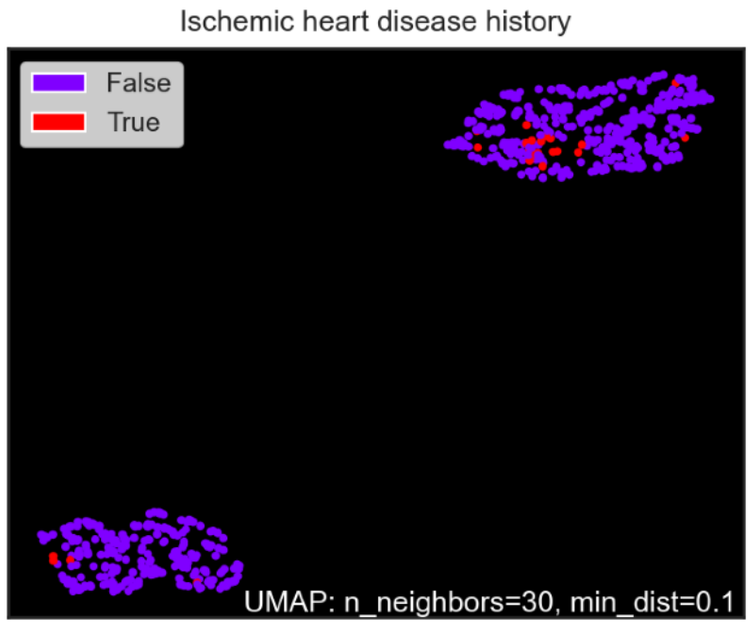
2+ aneurysms ?

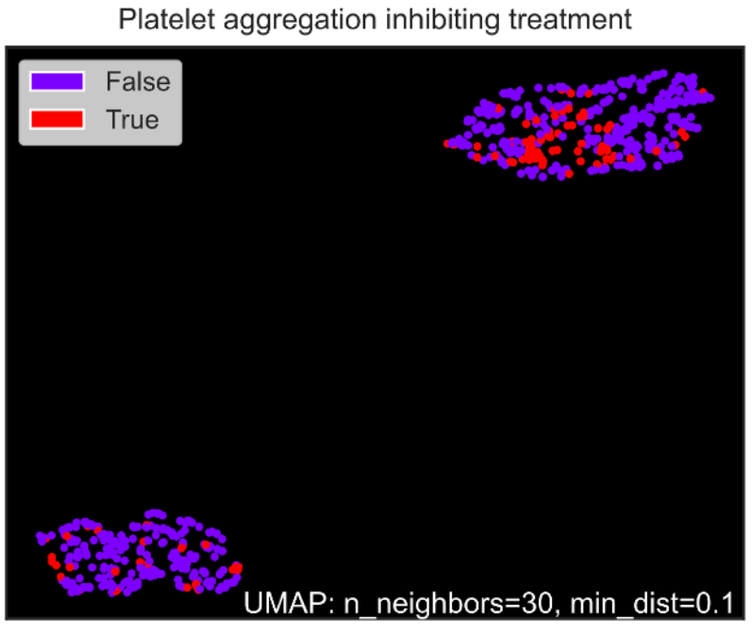
familial forms ?



Train machine-learning models ...







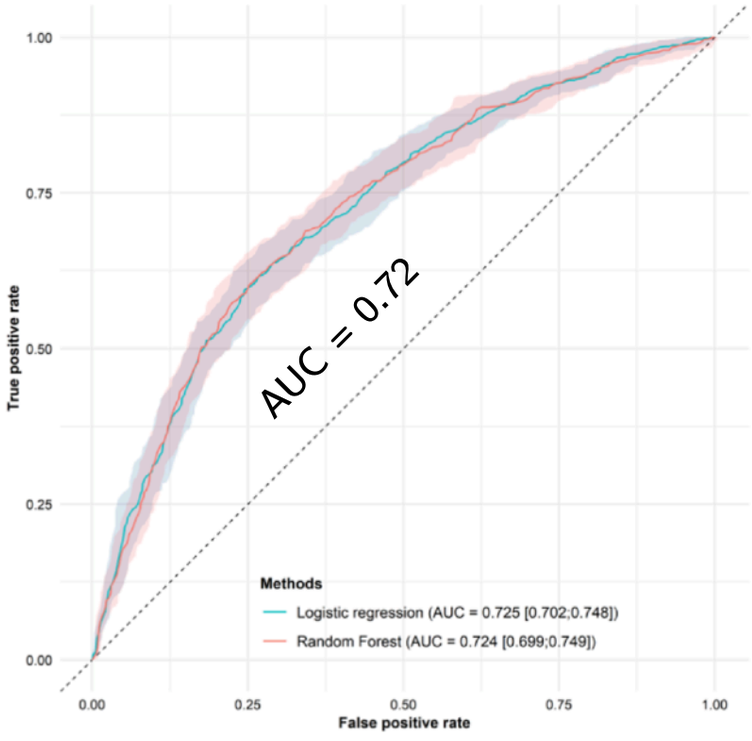
Observational Study > J Neurol Neurosurg Psychiatry. 2021 Feb;92(2):122-128.
doi: 10.1136/jnnp-2020-324371. Epub 2020 Oct 23.

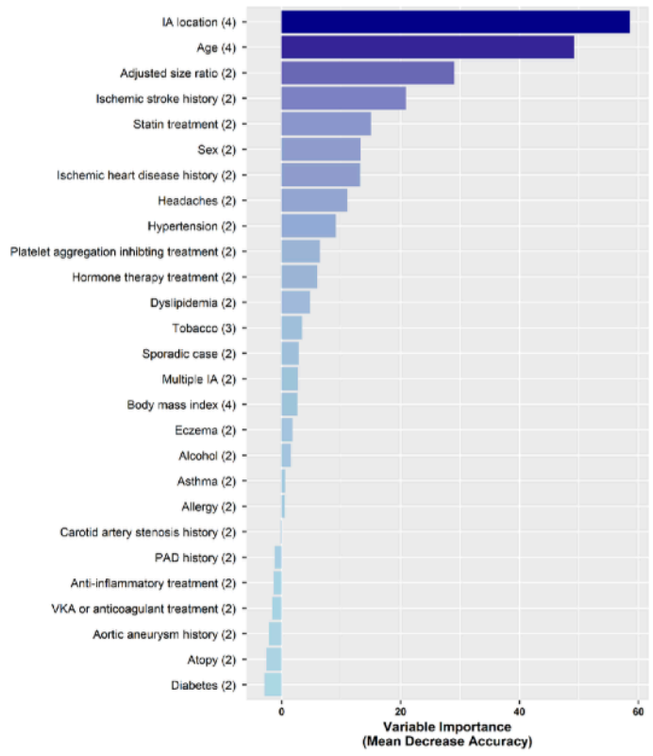
Location of intracranial aneurysms is the main factor associated with rupture in the ICAN population

Olivia Rousseau [1], Matilde Karakachoff [1], Alban Gaignard [2], Lise Bellanger [3], Philippe Bijlenga [4], Pacôme Constant Dit Beaufils [1], Vincent L'Allinec [5], Olivier Levrier [6], Pierre Aguettaz [7], Jean-Philippe Desilles [8], Caterina Michelozzi [9], Gaultier Marnat [10], Anne-Clémence Vion [2], Gervaise Loirand [2], Hubert Desal [11], Richard Redon [2], Pierre-Antoine Gourraud [1], Romain Bourcier [12]; ICAN Investigators

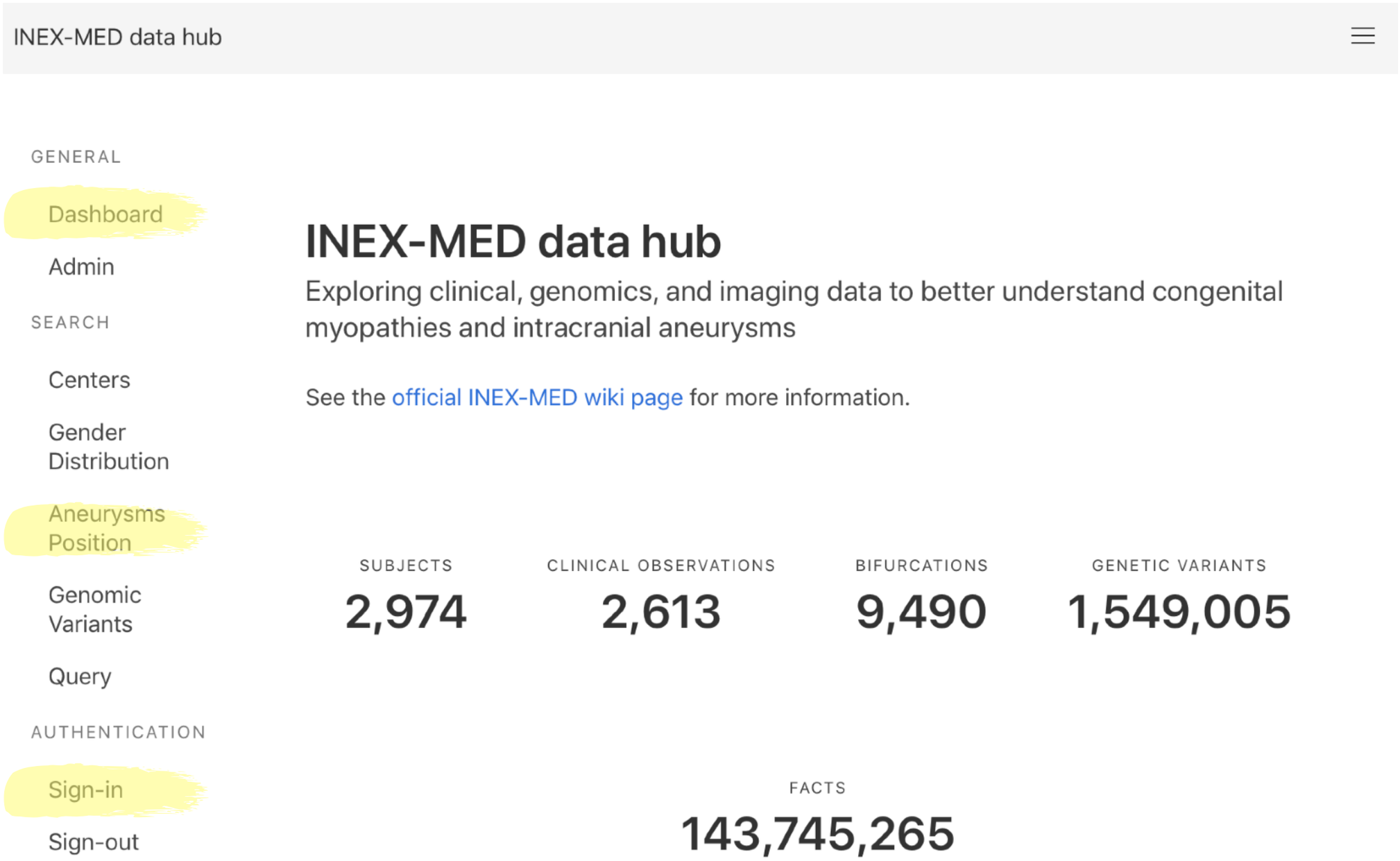
Collaborators, Affiliations + expand

PMID: 33097563 DOI: 10.1136/jnnp-2020-324371





(controlled) access and sharing



Web demonstrator → https://inexmed-api.univ-nantes.fr

Related works

Unruptured IA: 51 "core" common data elements / 1254

Age value, Birth date, Birth sex assigned type, Body system category, Race USA category, Sex genotype type, Ethnicity USA category, Gender identity type, Gender type,

Aneurysm diameter maximum measurement, Aneurysm height measurement, Aneurysm morphology type, Aneurysm width measurement,

Hypertension indicator,

Tobacco current use indicator, Tobacco prior use indicator, Tobacco use started age value, Tobacco use stopped age value,

World Federation of Neurological Surgeons (WFNS) - grading system subarachnoid hemorrhage scale

Behavioral history assessment date and time,

Biospecimen collection other text, Biospecimen collection type,

Cerebrospinal fluid collection anatomic site, Cerebrospinal fluid collection anatomic site other text, Cerebrospinal fluid collection method other text, Cerebrospinal fluid collection method type,

Data collected date and time, Data source,

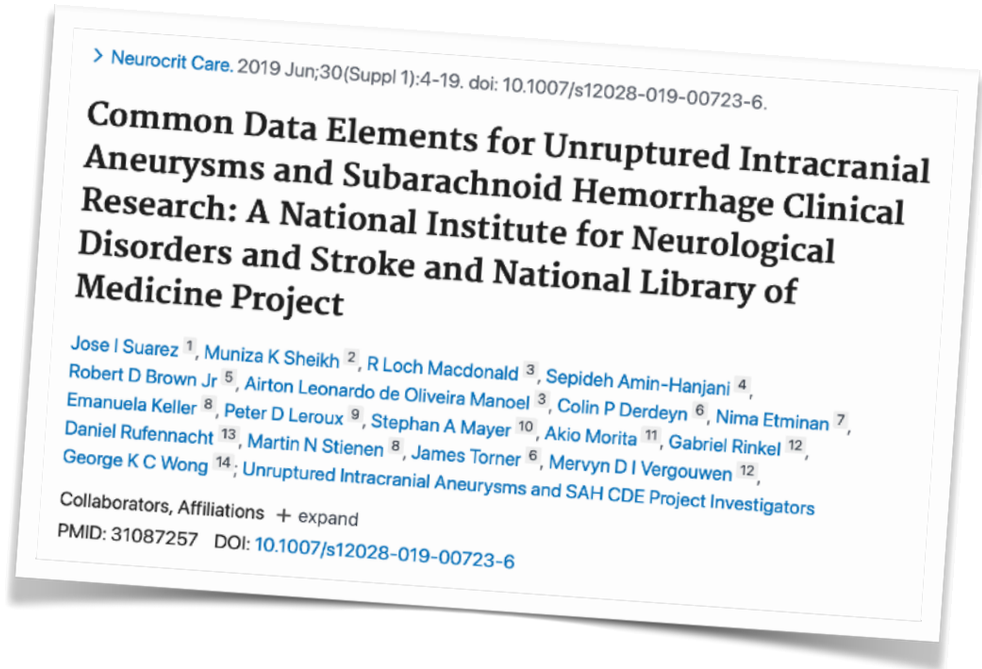
History data not obtained reason,

Imaging modality other text, Imaging modality type, Imaging modality vessel imaging angiography type, Imaging vessel angiography arterial anatomic site, Imaging vessel angiography arterial result,

Intervention endovascular status, Intervention surgical status, Lab specimen collection date and time,

Medical history condition SNOMED CT code, Medical history condition text, Medical history taken date and time,

Specimen acquisition anatomical site, Specimen baseline collection indicator, Specimen biologic source type, Specimen centrifugation duration, Specimen centrifugation revolutions per minute value, Specimen centrifugation temperature value, Specimen collection tube other text, Specimen collection tube type, Specimen not collected reason, Specimen storage temperature value, Specimen time collection process interval,



> Neurocrit Care. 2019 Jun;30(Suppl 1):4-19. doi: 10.1007/s12028-019-00723-6.

Common Data Elements for Unruptured Intracranial Aneurysms and Subarachnoid Hemorrhage Clinical Research: A National Institute for Neurological Disorders and Stroke and National Library of Medicine Project

Jose I Suarez [1], Muniza K Sheikh [2], R Loch Macdonald [3], Sepideh Amin-Hanjani [4], Robert D Brown Jr [5], Airton Leonardo de Oliveira Manoel [3], Colin P Derdeyn [6], Nima Etminan [7], Emanuela Keller [8], Peter D Leroux [9], Stephan A Mayer [10], Akio Morita [11], Gabriel Rinkel [12], Daniel Rufennacht [13], Martin N Stienen [8], James Torner [6], Mervyn D I Vergouwen [12], George K C Wong [14]; Unruptured Intracranial Aneurysms and SAH CDE Project Investigators

Collaborators, Affiliations + expand

PMID: 31087257 DOI: 10.1007/s12028-019-00723-6

- ✓ Large community effort
- ✓ Consensual definitions
- ⚠ Flat list of terms
- ⚠ No recommendation data structures

Sample knowledge graphs : SPHN network

nature > scientific data > articles > article

Article | Open Access | Published: 10 March 2023

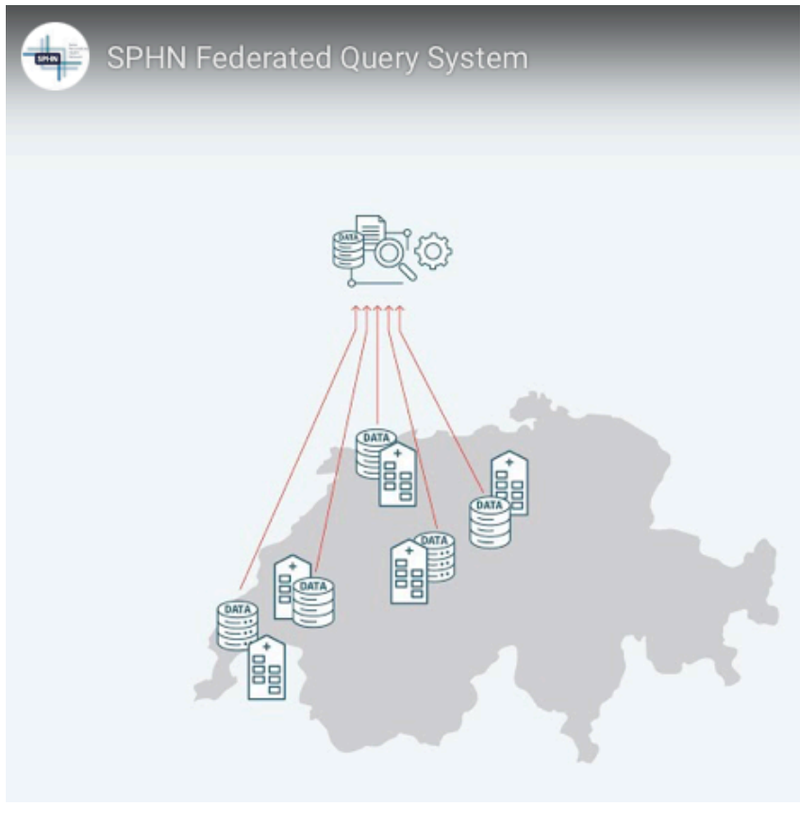
FAIRification of health-related data using semantic web technologies in the Swiss Personalized Health Network

Vasundra Touré, Philip Krauss, Kristin Gnodtke, Jascha Buchhorn, Deepak Unni, Petar Horki, Jean Louis Raisaro, Katie Kalt, Daniel Teixeira, Katrin Crameri & Sabine Österle

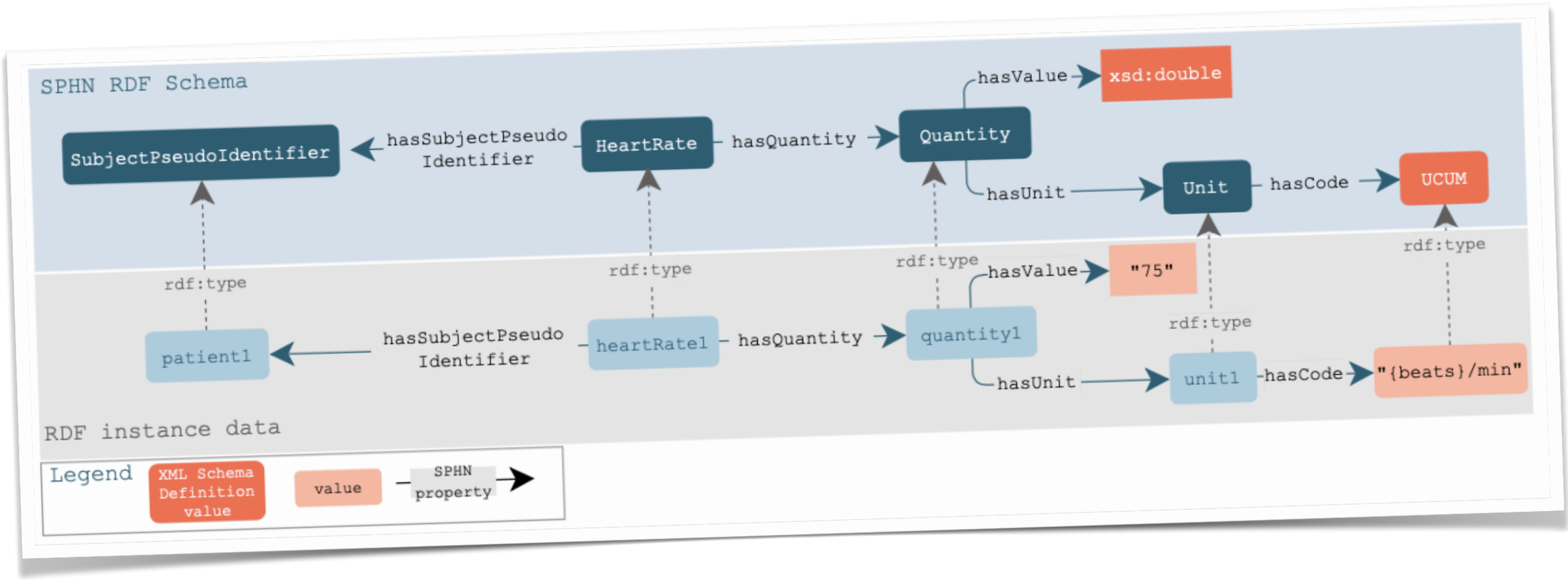
Scientific Data **10**, Article number: 127 (2023) | Cite this article

Abstract

The Swiss Personalized Health Network (SPHN) is a government-funded initiative developing federated infrastructures for a responsible and efficient secondary use of health data for research purposes in compliance with the FAIR principles (Findable, Accessible, Interoperable and Reusable). We built a common standard infrastructure with



Sample knowledge graphs : SPHN network



+ Allergy
+ Biobank
+ Consent
+ Radiology diagnosis
+ Genomic variants position
+ ECG

Sample knowledge graphs: Malik et. al. 2020

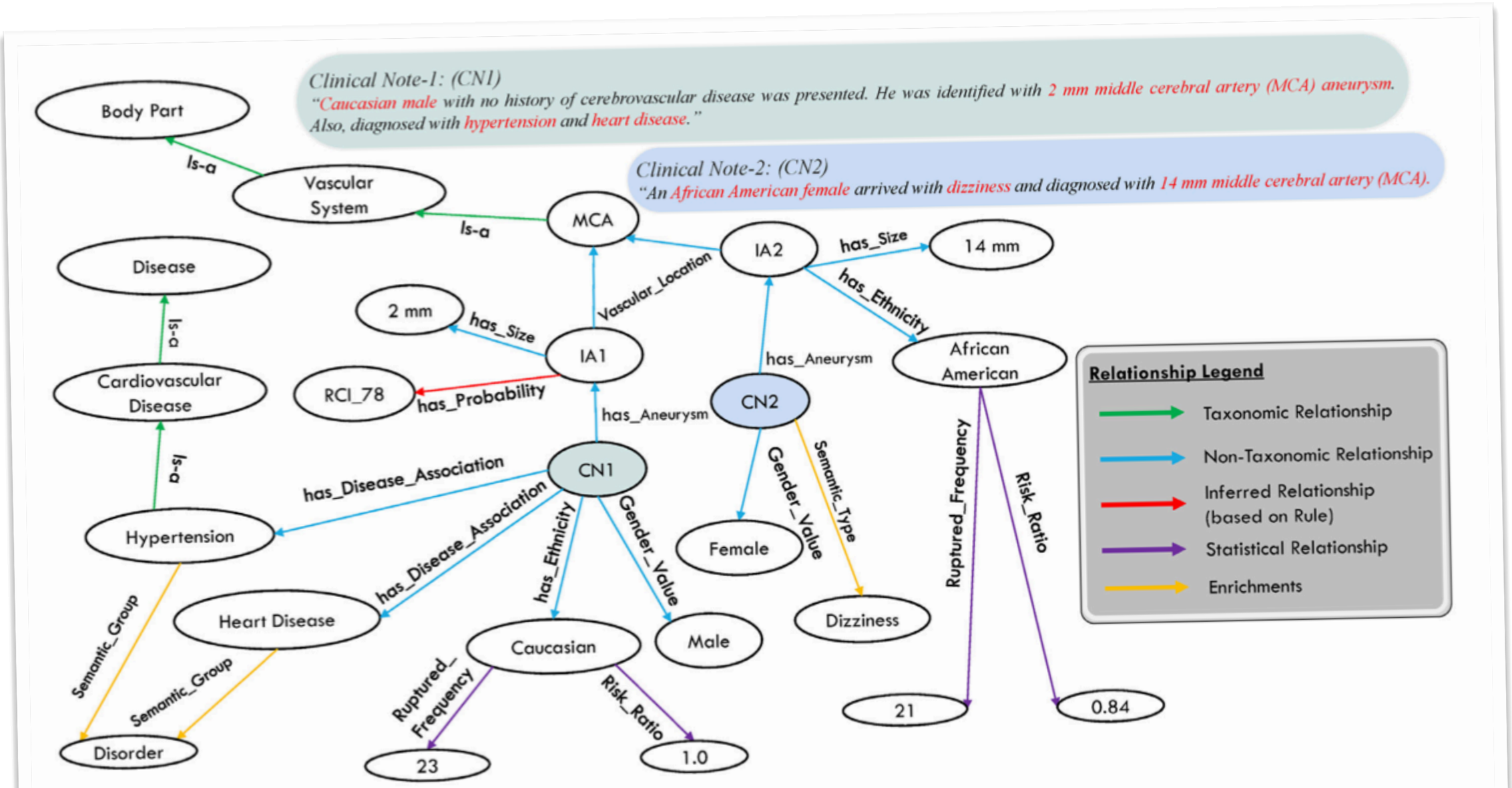


Fig. 3. An excerpt/sub-graph of saccular aneurysm KG of middle cerebral artery defining IA risk factors based on two clinical texts.

Malik, K. M., Krishnamurthy, M., Alobaidi, M., Hussain, M., Alam, F., & Malik, G. (2020). ***Automated domain-specific healthcare knowledge graph curation framework: Subarachnoid hemorrhage as phenotype****. Expert Systems with Applications, 145, 113120.*

Future works

Wrap up & next steps

- Gather **consensual expert knowledge** with computational **Ontologies**
- Knowledge Graphs are "**machine-processable**"
- Link **multi-scale data** with Knowledge Graphs
- Query **multi-source** data

PEPR Santé numérique

- Include familial relations to query IA **familial forms**
- Represent **care pathways and guidelines**
- Develop a national **data hub** with FAIR intracranial aneurysms data
- **Share** and enrich data models with **international** initiatives, *but which ones ?*
- Further develop **predictive approaches** (rupture risk prediction)

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Olivia Rousseau,
Hala Skaf-Molli,
Kirsley Chennen,
Jocelyn Laporte,
Olivier Poch,
Norma Romero,
Julie Thompson

ICAN consortium

BiRD facility,
NeurInfo facility