SIBioC Medicina di Laboratorio ETS

AI in medicina di laboratorio: stato dell'arte e prospettive

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CRO AVIANO IRCCS CRO Istituto di ricovero e cura a carattere scientifico "Centro di Riferimento Oncologico" di Aviano

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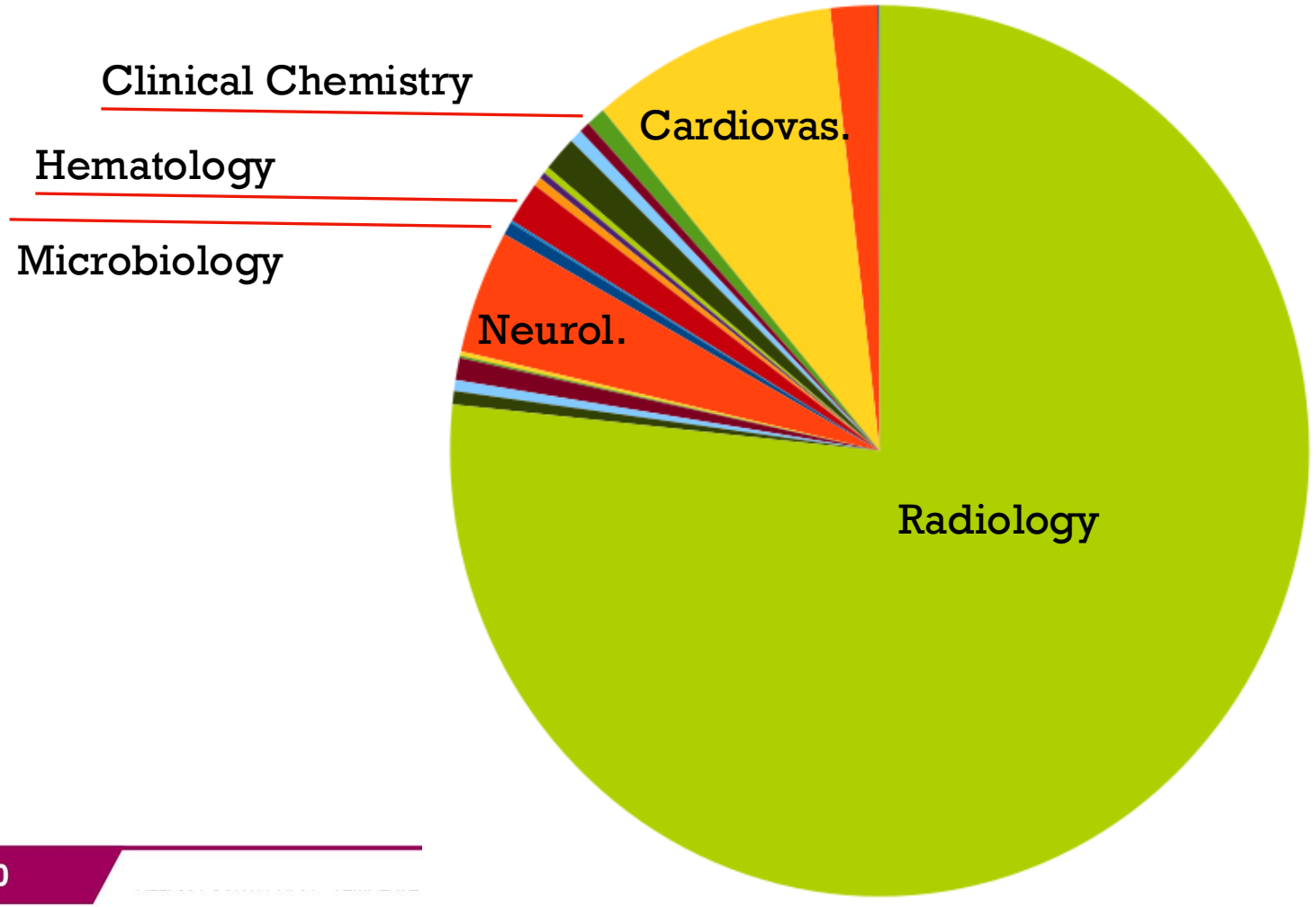
EFLM EUROPEAN FEDERATION OF CLINICAL CHEMISTRY AND LABORATORY MEDICINE

ifcc International Federation of Clinical Chemistry and Laboratory Medicine

U.S. FOOD & DRUG ADMINISTRATION

AI-Enabled Medical Devices List



Clinical importance of laboratory data

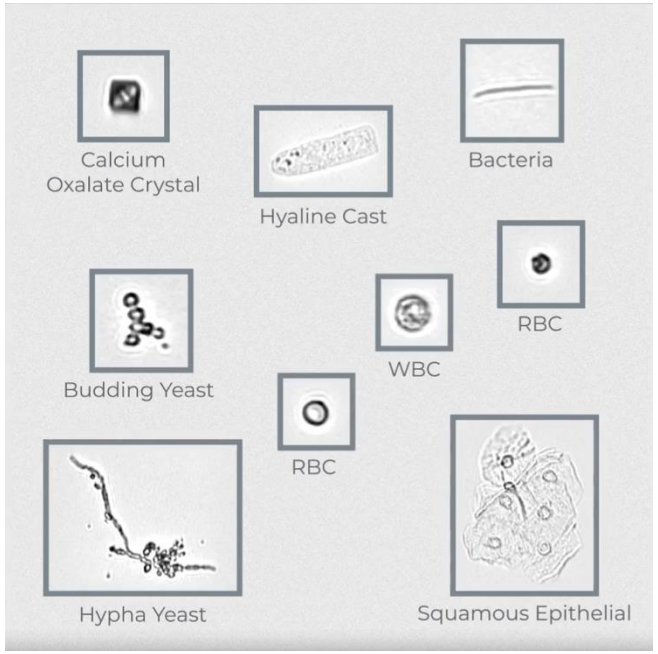
- "70% of clinical decisions guided by laboratory data"
- "5% of total costs affecting 95% of other costs"

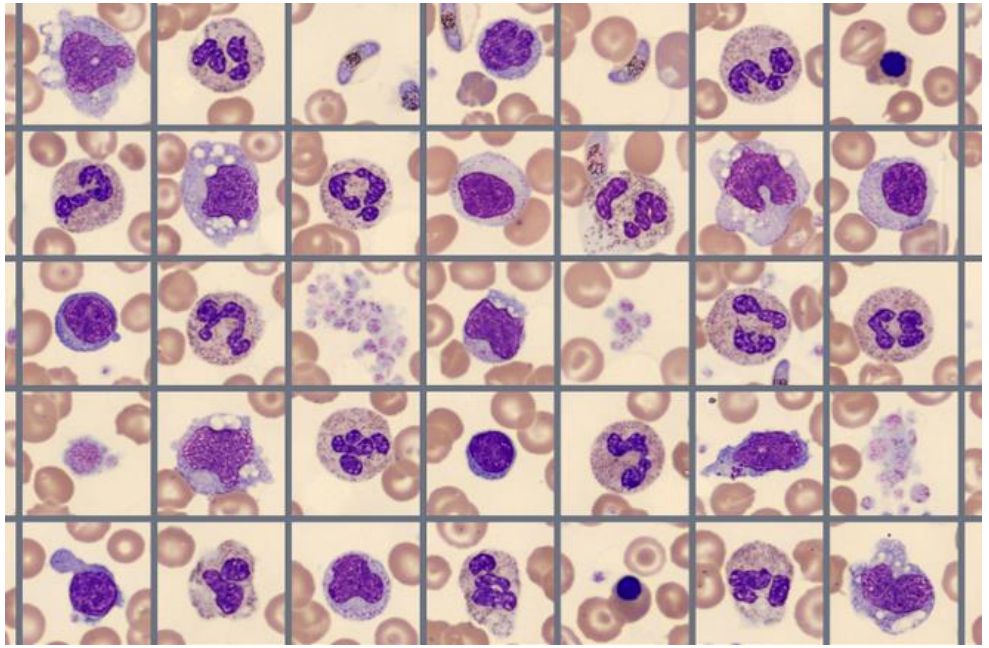
Hallworth, M. J. The '70% claim': what is the evidence base? *Ann Clin Biochem* **48**, 487–488 (2011).

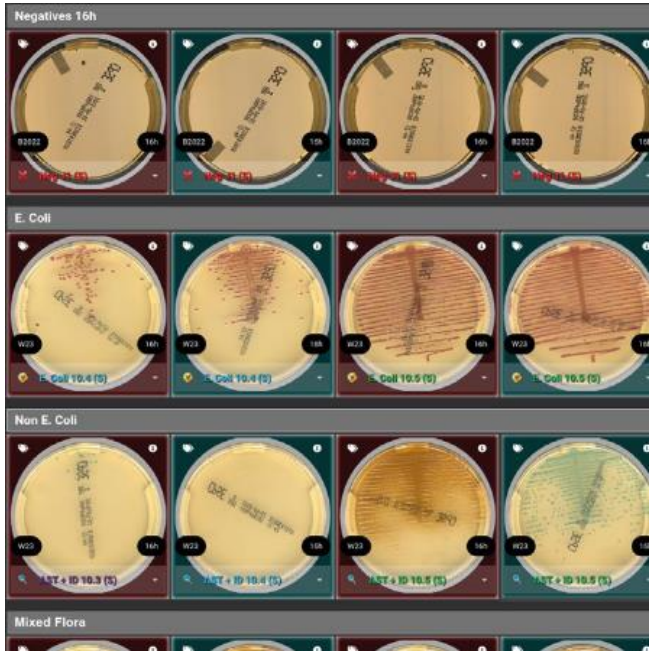
- 1.4-2.3% of costs and 66% of decisions (50% if considering 75% of patients undergoing IVD).

Rohr, U.-P. *et al.* The Value of In Vitro Diagnostic Testing in Medical Practice: A Status Report. *PLOS ONE* **11**, e0149856 (2016).

Main types of AI-based systems in the lab







FDA U.S. FOOD & DRUG ADMINISTRATION

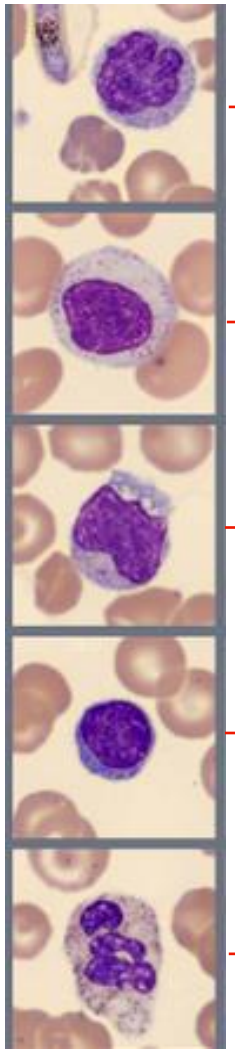
AI-Enabled Medical Devices List

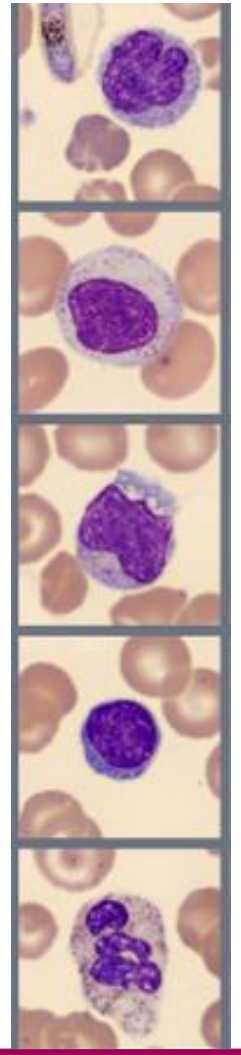
DeNovo: Low/moderate risk new devices

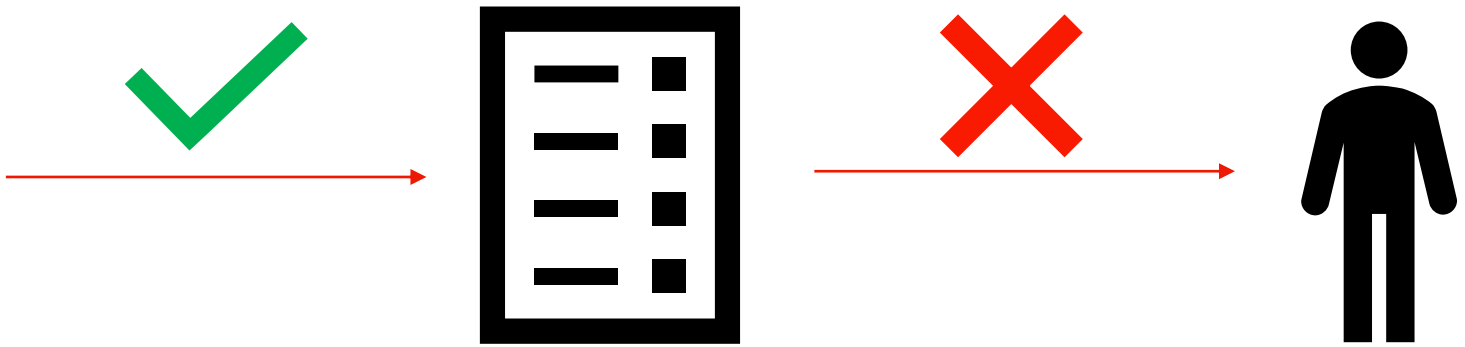
Date	#	Product	Company	Panel	Description
06/29/2023	DEN200052	KidneyIntelX.dkd	[redacted]	Clinical Chemistry	Scoring for progressive renal disease (on immunoassay)

Features of FDA-approved clinical laboratory applications

- Mainly Image-based
- Classification support
  - They answer the question: What is this object?
- "Technical" applications
  - Data --> technical information
  - Focused on efficiency - not adding clinical insights





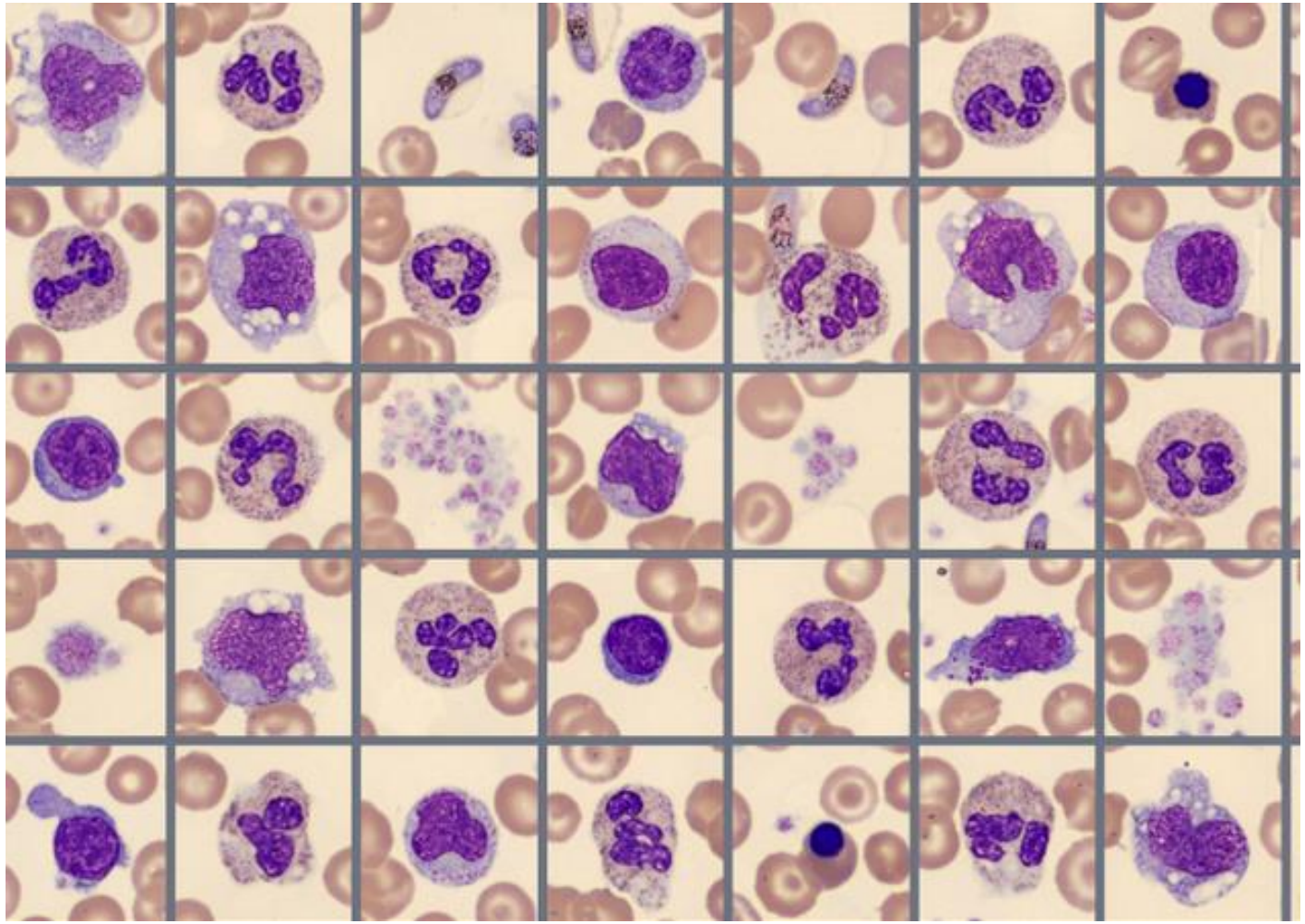


- Insight on clinical outcome
- Data --> "clinical" information
- Focus on effectiveness
- Impacting on patient outcome

Questions

Why "technical" lab applications and not "clinical"?

Why mainly image-based applications?



Images

- **Need for human classification**
  - Image is not readable by everyone
- Image is largely context-independent
- Decision support of what is already known
  - Final validation is human
  - Human can easily check if AI was right

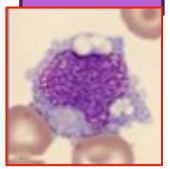
"Easily" validated tools

Narrow: executing a specific and isolated task

Interpretable/verifiable: showing reasons on which the output is based or can be easily verified

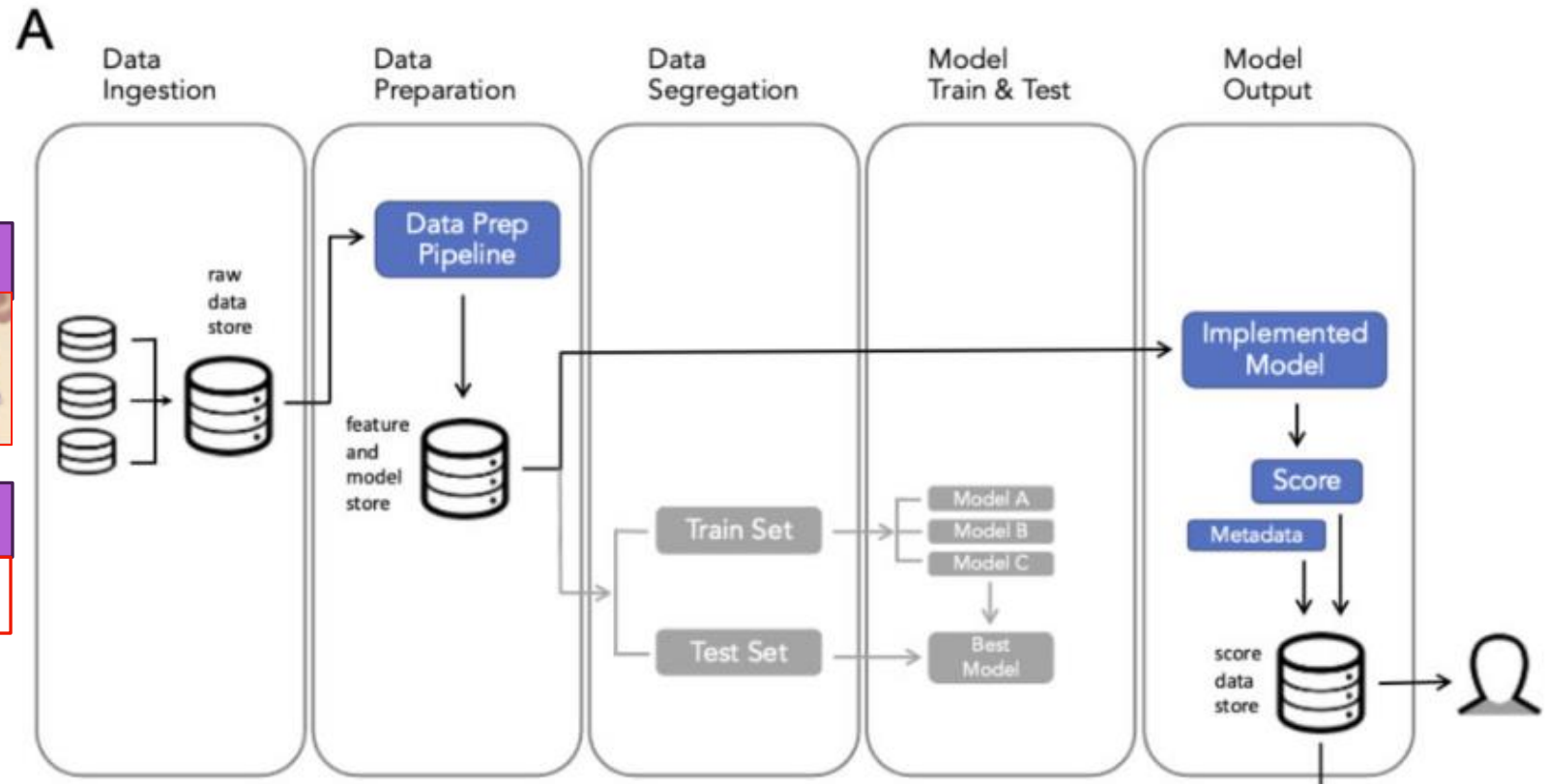
Pipeline of a "technical" machine learning model

Data



Tech. valid information

Monocyte



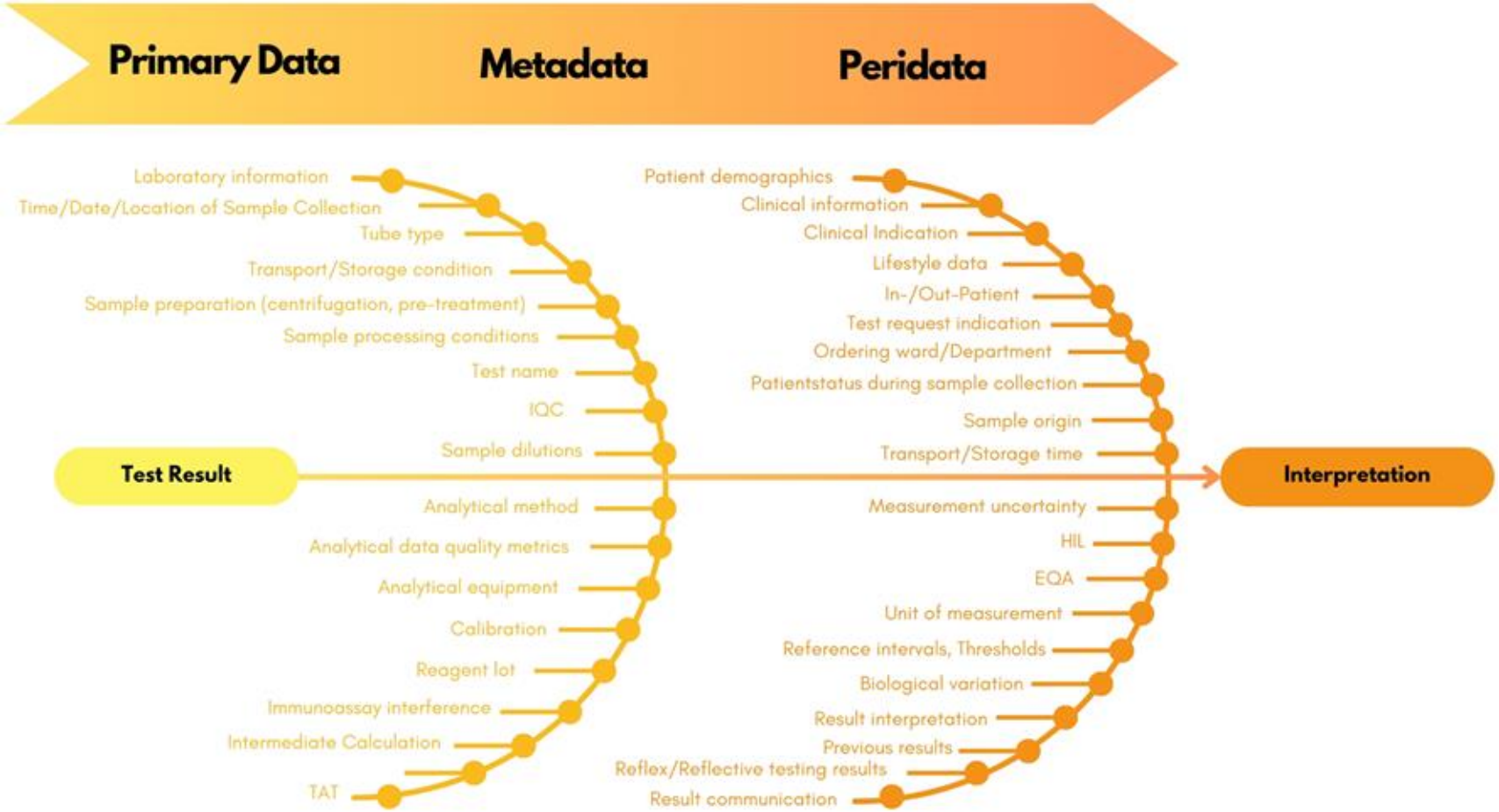
Taken from Haymond, Shannon and Christopher McCudden. "Rise of the Machines: Artificial Intelligence and the Clinical Laboratory." J. Appl. Lab. Med., vol. 6, no. 6, 1 Nov. 2021, pp. 1640-54, doi:10.1093/jalm/jfab075.

Why **limited** applications on laboratory values?

Numbers are ideal inputs of machine learning models

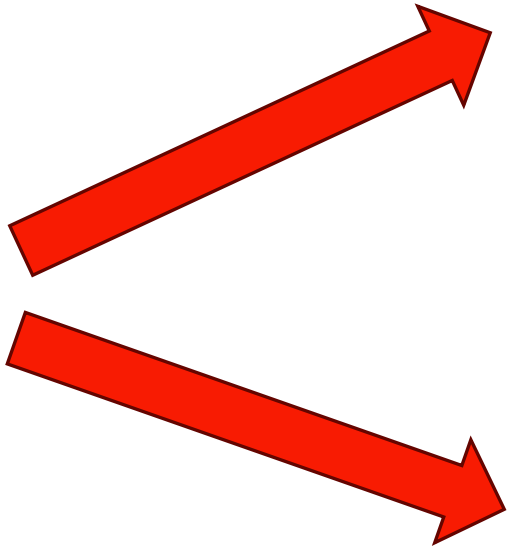
The apparent simplicity of the laboratory value

- No need for human classification
  - A number is (apparently) readable by a clinician
  - A number with reference interval is (apparently) readable by everyone



Padoan, A. et al. "Data flow in clinical laboratories: could metadata and peridata bridge the gap to new AI-based applications?: An investigation on behalf of the European Federation of Clinical Chemistry and Laboratory Medicine (EFLM) Working Group on Artificial Intelligence (WG-AI)" *Clinical Chemistry and Laboratory Medicine (CCLM)*, vol. 63, no. 4, 2025, pp. 684-691.

Hypoglycemia



Because of the disease?

Because blood draw was performed 8 hours before?

The apparent simplicity of the laboratory value

- Laboratory values are heavily **context-dependent**
  - Interpretation depends on **time patterns + combination of lab values + age/gender + clinical data** → need for multimodal AI
  - AI on laboratory data **needs to be integrated with EHR and clinical context**, making training, validation and implementation more complex

Carobene A, Cadamuro J, Frans G, et al. EFLM checklist for the assessment of AI/ML studies in laboratory medicine: enhancing general medical AI frameworks for laboratory-specific applications. *Clin Chem Lab Med, 2025.*

Pipeline of a "clinical" machine learning model

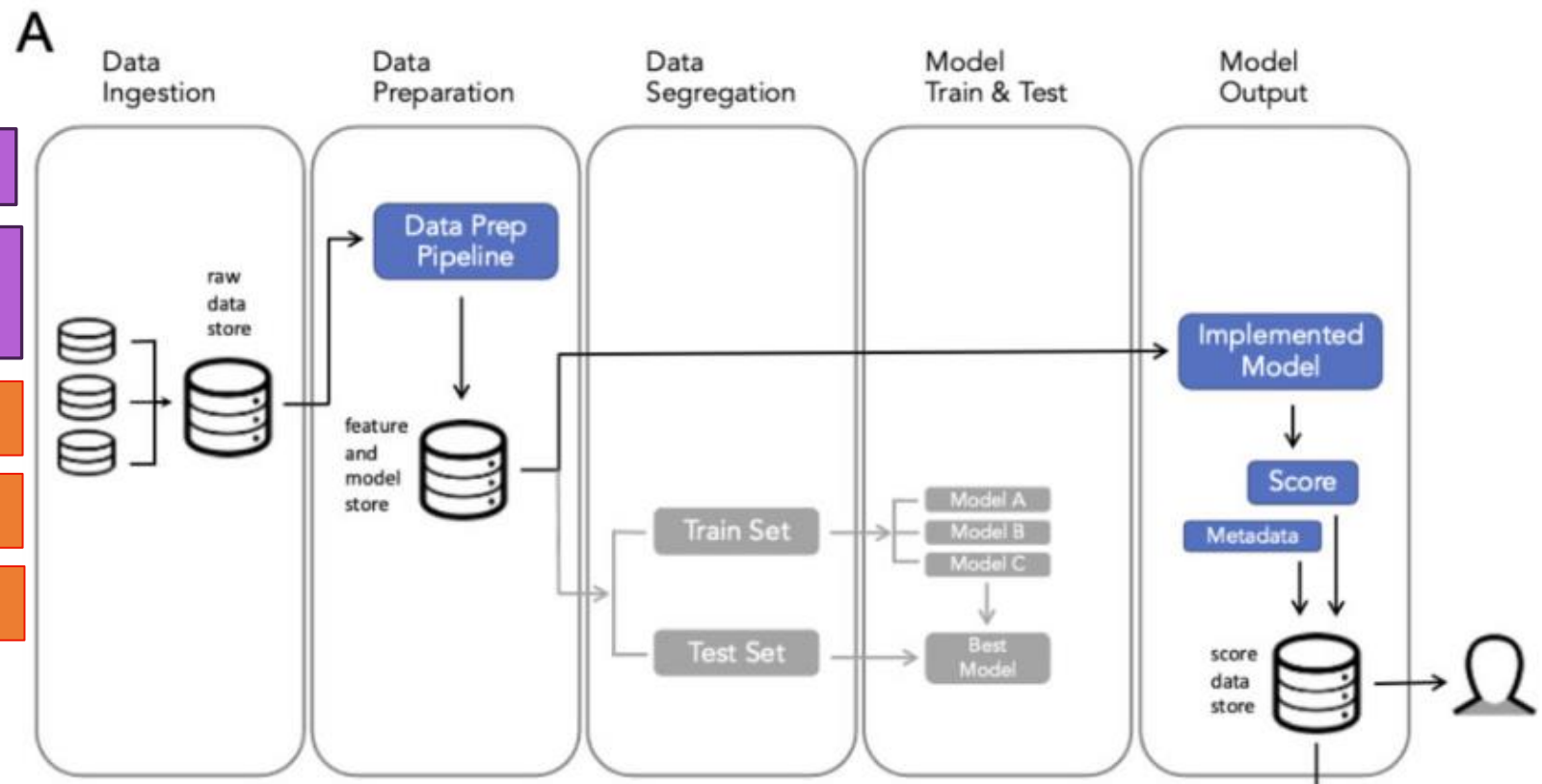
Data

Tech/clin. valid information

Meta/Peri Data

Clinical information (EHR)

Clinical outcomes (EHR)



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Lack of Meta/PeriData information

Despite many data are digitally present, most of them are not recorded by LIS in the digital archive nor in EHR/FSE.

Laboratory data Clinical outcomes

- Today mostly manual
  - Slow
  - Labour-intensive
  - Cost-ineffective
  - Dangerous! – Students accessing patient data

Proof of failure

PRISM – Precision Retrospective Integrative Study for Metastatic lymph nodes in Breast Cancer

- Problem: Sentinel lymph node biopsy is invasive and 70% negative
- Solution: Replace lymph node biopsy with non-invasive technique
- Explored implementation: MRI |> AI
- Proposed Implementation: MRI + Lab data + Clinical data |> AI

Proof of failure

PRISM – Precision Retrospective Integrative Study for Metastatic lymph nodes in Breast Cancer

- Difference in approval time between EU countries
  - (1 month Austria vs 1.5 year Italy)
- Austria database was incomplete
  - Clinical outcome not accessible (different database)
- Italy database 20% progress in upload after 2 years (manual upload)

What would it be a game changer?

Automatic controlled access to clinical outcomes

No additional efforts by clinicians: Maintaining unstructured clinical reports

Tool for systematic access of healthcare data

Effective methods to structure the unstructured clinical notes

LLM are extremely effective in data extraction from unstructured EHR

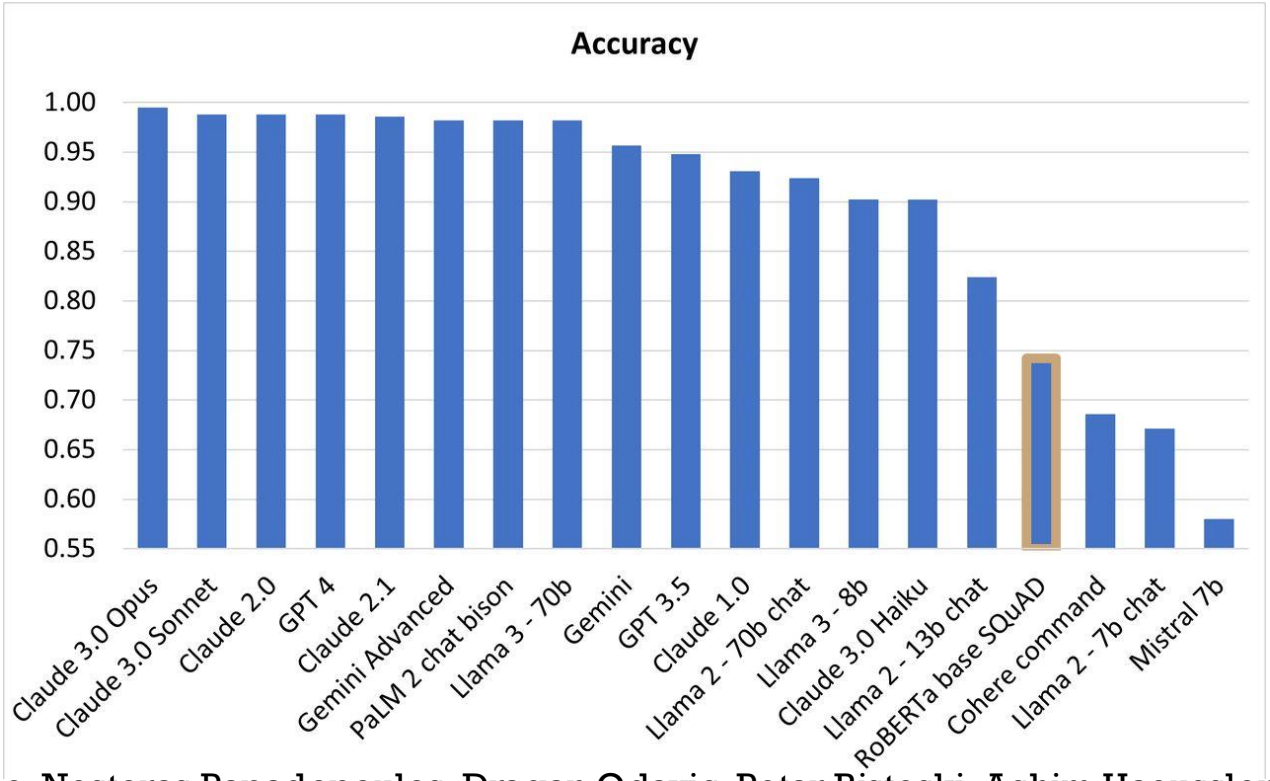
Open access Original research

BMJ Health & Care Informatics

Large language models for data extraction from unstructured and semi-structured electronic health records: a multiple model performance evaluation

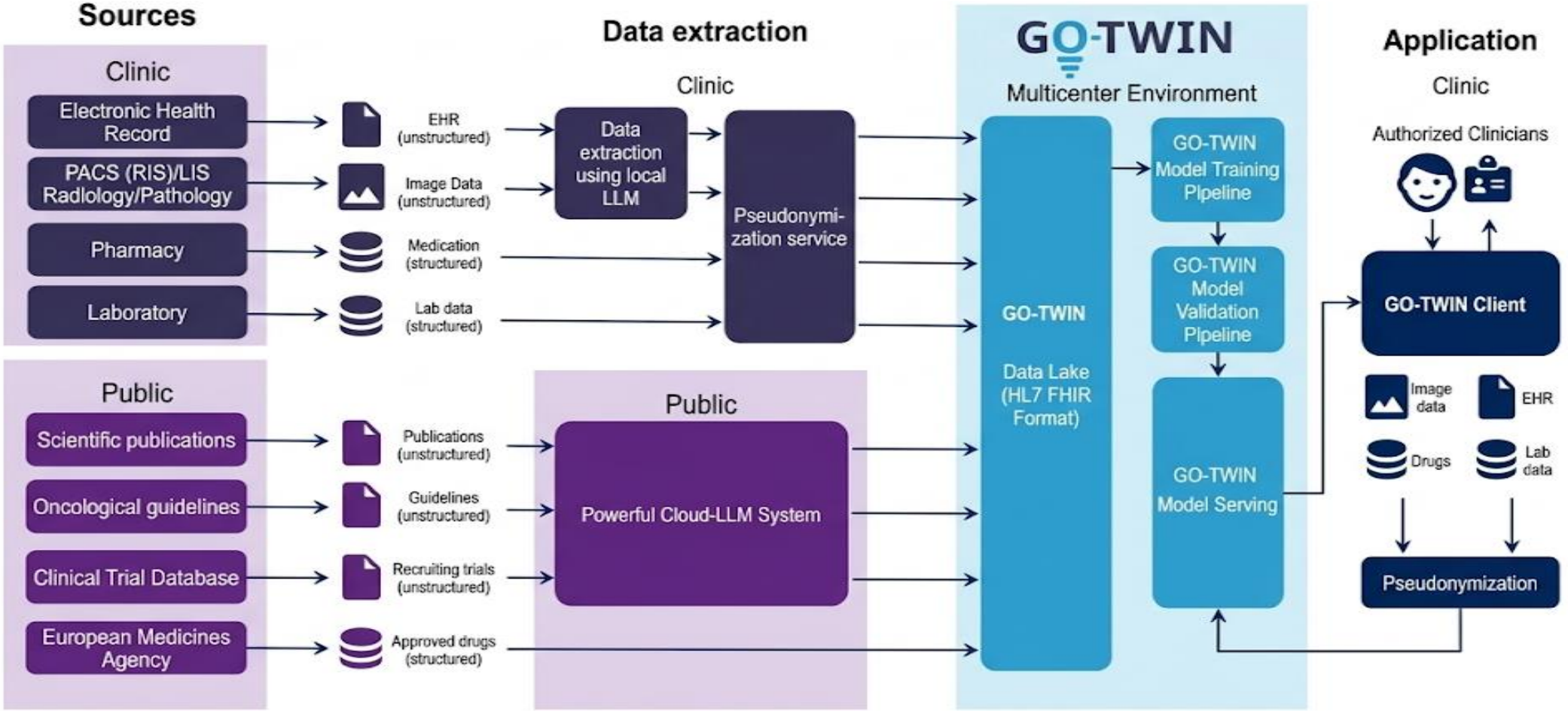
Vasileios Ntinopoulos [1,2] Hector Rodriguez Cetina Biefer,[1,2] Igor Tudorache,[1,2] Nestoras Papadopoulos,[1,2] Dragan Odavic,[1,2] Petar Risteski,[1,2] Achim Haeussler,[1,2] Omer Dzemali [1,2]

- 8 models shown accuracy > 98%
- Baseline model accuracy = 74%



Vasileios Ntinopoulos, Hector Rodriguez Cetina Biefer, Igor Tudorache, Nestoras Papadopoulos, Dragan Odavic, Petar Risteski, Achim Haeussler, Omer Dzemali - Large language models for data extraction from unstructured and semi-structured electronic health records: a multiple model performance evaluation: BMJ Health & Care Informatics 2025;32:e101139.

GO-TWIN architecture enables privacy-compliant, multimodal AI applications



Lammert J, et al. Large language models-enabled digital twins for precision medicine in rare gynecological tumors. *npj Digital Medicine*. 2025;8(1). doi:10.1038/s41746-025-01810-z

Interreg Co-funded by the European Union

AI-Brain project

Locally deployed AI to structure clinical notes for effective biobanking

ASU FC Azienda sanitaria universitaria Friuli Centrale

CRO AVIANO

MEDIZINISCHE UNIVERSITÄT INNSBRUCK

Fraunhofer AUSTRIA

SRL FVG

GPDP GARANTE PER LA PROTEZIONE DEI DATI PERSONALI

FSE – finalità di ricerca solo per alcuni enti

Regioni e province autonome, del Ministero del lavoro e delle politiche sociali, del Ministero della salute e dall'Agenzia nazionale per i servizi sanitari regionali (AGENAS)

EDS – Ecosistema Dati Sanitari – disponibile agli enti tramite richiesta ad AGENAS

Non disponibile in questo momento. Previsto entro il 31/12/2026 (Decreto del Ministero della Salute del 31 dicembre 2024)

Modalità della richiesta e tempi di evasione? Modifica della richiesta in tempo reale? Rischio silos burocratico centrale? Rischio di limitata transizione digitale sul territorio?

SIBioC
Medicina di Laboratorio ETS

Il laboratorio necessita di un'interfaccia di accesso diretto con profilo specifico all'EDS per poter efficacemente eseguire:

1. **Monitoraggio del valore clinico erogato (*Value-Based Laboratory Medicine*)**
2. **Ottimizzazione dell'appropriatezza prescrittiva**
3. **Ricerca predittiva e scoperta di nuovi biomarcatori**

1. **Monitoraggio del valore clinico erogato (*Value-Based Laboratory Medicine*)**

- **Es.: Quanti malattie Y si sono diagnosticate rispetto al totale delle richieste dell'esame X?**
- **Quanti giorni di ricovero ho evitato da quando ho introdotto l'esame Z?**

Due parole sul value-based: Laboratory value = Outcome/Cost

E' fuorviante valorizzare il laboratorio sul volume delle prestazioni

Science

RESEARCH ARTICLE

Dissecting racial bias in an algorithm used to manage the health of populations

ZIAD OBERMEYER, BRIAN POWERS, CHRISTINE VOGELI, AND SENDHIL MULLAINATHAN Authors Info & Affiliations

The bias arised because the algorithm **predicted health care costs rather than illness…**

Obermeyer Z, Powers B, Vogeli C, Mullainathan S. Dissecting racial bias in an algorithm used to manage the health of populations. *Science*. 2019;366(6464):447-453. doi:10.1126/science.aax2342

2. Ottimizzazione dell'appropriatezza prescrittiva

- es.: La rimozione dell'esame X ha impattato sulle diagnosi di malattia Y?

Due parole sull'appropriatezza:

Assicura la sostenibilità

Limita la completezza dei database, e di conseguenza la scoperta di relazioni inattese

Se chiediamo solo ciò che è noto, sarà difficile scoprire l'ignoto

3. Ricerca predittiva e scoperta di nuovi biomarcatori

- **Es.: Ci sono combinazioni di valori dell'emocromo che possono predire una patologia X?**

Take home messages

Keys to unlock the full potential of AI in laboratory medicine

- Integrating Metadata and Peridata into EHR
  - Hidden layer?
- Tools for systematic secure access to EHR
  - Ecosistema dei Dati Sanitari? Decreto del Ministero della Salute del 31 dicembre 2024 – AGENAS entro il 31/12/2026
- Use of effective systems to structure clinical notes
  - Local LLM?

Grazie per l'attenzione

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