PERSONALIZED MEDICINE IN THE ARTIFICIAL INTELLIGENCE ERA

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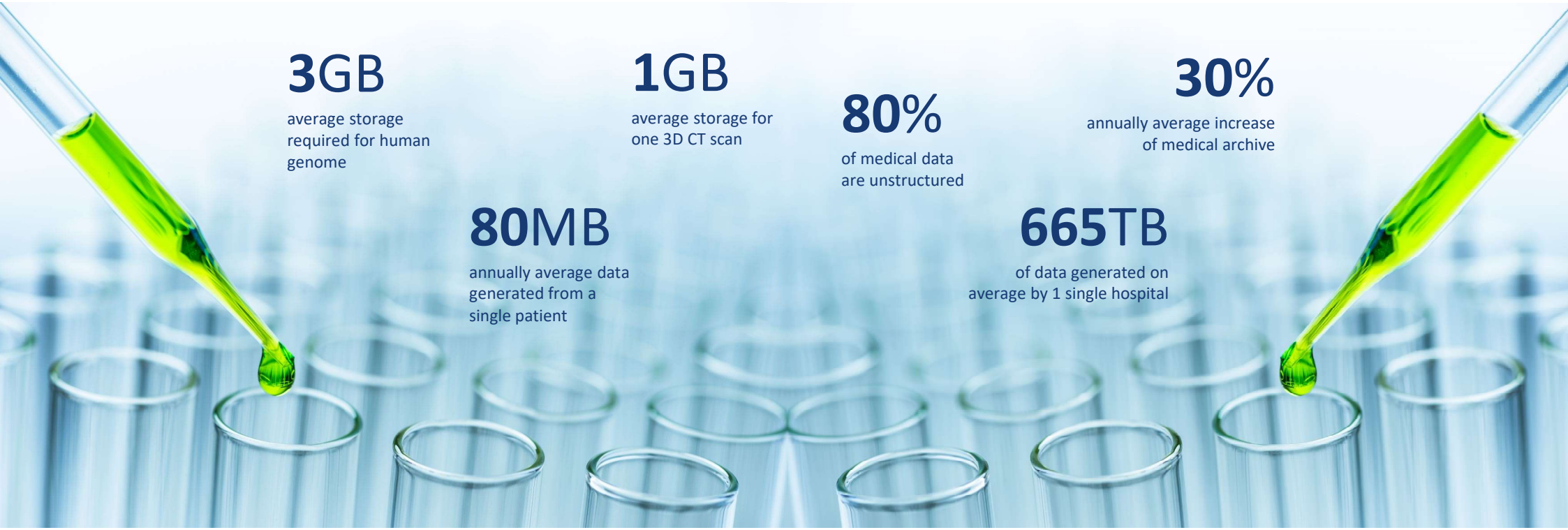
HEALTHCARE BIGDATA: VOLUME, VELOCITY AND VARIETY



3GB
average storage required for human genome

1GB
average storage for one 3D CT scan

80%
of medical data are unstructured

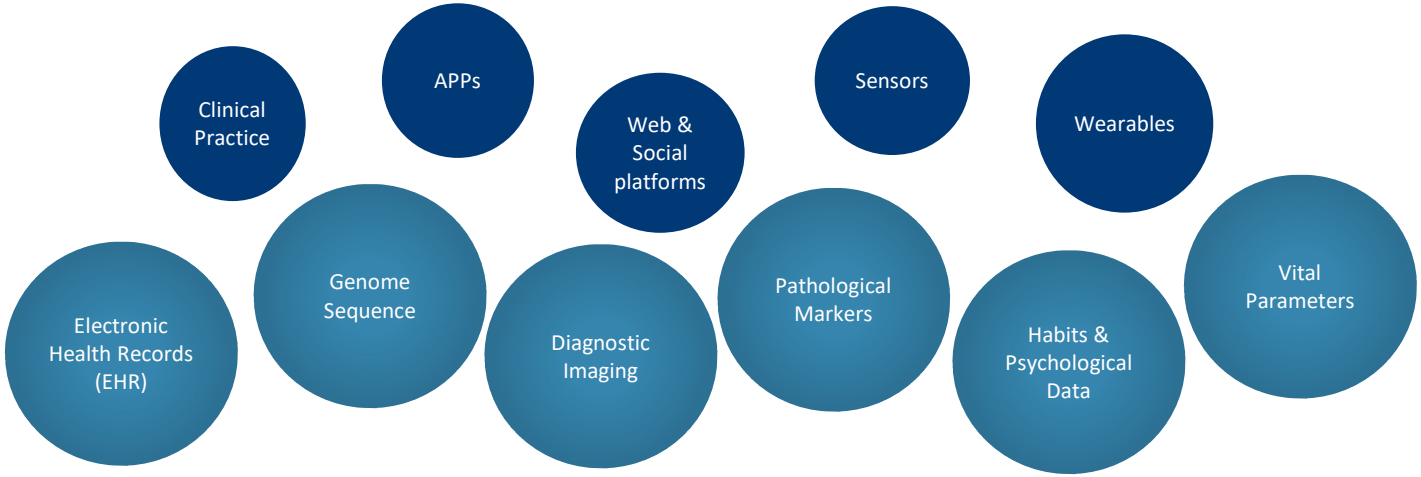
30%
annually average increase of medical archive

80MB
annually average data generated from a single patient

665TB
of data generated on average by 1 single hospital

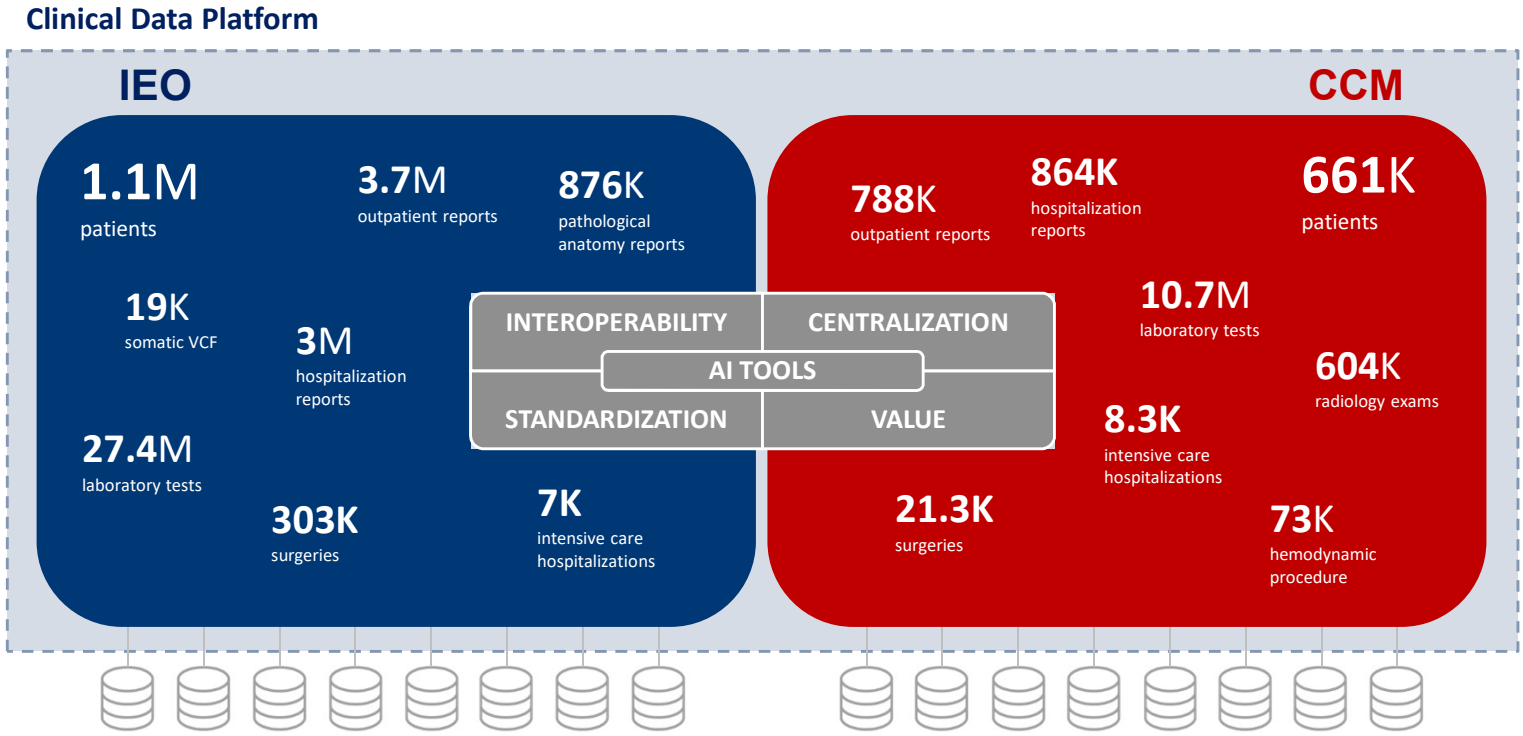
https://www.bigdata4health.unimore.it/bd4h/
https://storage.pardot.com/22172/10472/health_graph.jpg

DIGITAL HEALTH ENVIRONMENT: PATIENT DIGITALIZATION AND MULTI-STANDARDS



FHIR	SDA	HL7 v2	OMOP
IHE	CDA	DICOM	XML

IEO AND CCM SOLUTION: CLINICAL DATA PLATFORM



AI MODELS FOR PERSONALIZED MEDICINE

AI MODELS: THERAPIES

Clinical Data Platform

EHRs

THERAPIES

ID	Line of Therapy	Setting	Therapy	Start Date	End Date
1	1	Adjuvant	['FEC' per 3 , 'Docetaxel' per 3	26.10.2012	
1	2	Post-relapse adjuvant	['CMF'] per 6 cicli	08.09.2014	05.03.2015
1	3	II Post-relapse adjuvant	['ViFuP']	06.07.2015	02.09.2015
1	5	II Post-relapse adjuvant	['Ciclofosfamide', 'Methotrexate']	15.01.2016	
1	6	I line	['Caelyx']	14.03.2016	20.06.2016
1	7	II line	['Pembrolizumab']	04.08.2016	
1	8	II line	['Abraxane']	19.12.2016	29.05.2017
1	9	III line	['Capecitabina'] metronomica	19.06.2017	14.09.2017
1	10	IV line	['Eribulina']	18.09.2017	30.11.2017
1	11	V line	['Vinorelbina'] metronomica	28.11.2017	
2	1	Adjuvant	['Caelyx']	16.05.2014	14.11.2014
...					

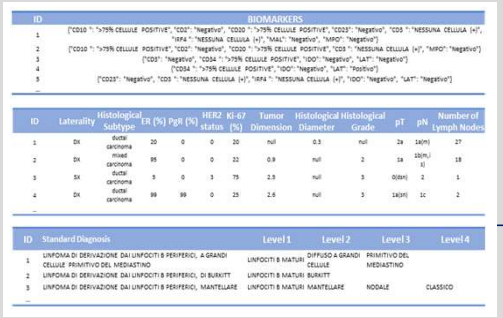
AI MODELS: PATHOLOGICAL DIAGNOSES & BIOMARKERS

Clinical Data Platform

Pathology Reports

BIOMARKERS

DIAGNOSES



ID	BIOMARKERS
1	{"CD10 ": ">75% CELLULE POSITIVE", "CD2": "Negativo", "CD20 ": ">75% CELLULE POSITIVE", "CD23": "Negativo", "CD5 ": "NESSUNA CELLULA (+)", "IRF4 ": "NESSUNA CELLULA (+)", "MALT": "Negativo", "MPO": "Negativo"}
2	{"CD10 ": ">75% CELLULE POSITIVE", "CD2": "Negativo", "CD20 ": ">75% CELLULE POSITIVE", "CD5 ": "NESSUNA CELLULA (+)", "MPO": "Negativo"}
3	{"CD3": "Negativo", "CD34 ": ">75% CELLULE POSITIVE", "IDO": "Negativo", "LAT": "Negativo"}
4	{"CD34 ": ">75% CELLULE POSITIVE", "IDO": "Negativo", "LAT": "Positivo"}
5	{"CD23": "Negativo", "CD5 ": "NESSUNA CELLULA (+)", "IRF4 ": "NESSUNA CELLULA (+)", "IDO": "Negativo", "LAT": "Negativo"}

ID	Laterality	Histological Subtype	ER (%)	PgR (%)	HER2 status	KI-67 (%)	Tumor Dimension	Histological Diameter	Histological Grade	pT	pN	Number of Lymph Nodes
1	DX	ductal carcinoma	20	0	0	20	null	0.5	null	2a	1a(mi)	27
2	DX	mixed carcinoma	95	0	0	22	0.9	null	2	1a	1b(mi, s)	18
3	SX	ductal carcinoma	5	0	3	75	2.5	null	3	0(is)	2	1
4	DX	ductal carcinoma	99	99	0	25	2.6	null	3	1a(sn)	1c	2

ID	Standard Diagnosis	Level 1	Level 2	Level 3	Level 4
1	LINFOMA DI DERIVAZIONE DAI LINFOCITI B PERIFERICI, A GRANDI CELLULE PRIMITIVO DEL MEDIASTINO	LINFOCITI B MATURI	DIFFUSO A GRANDI CELLULE	PRIMITIVO DEL MEDIASTINO	
2	LINFOMA DI DERIVAZIONE DAI LINFOCITI B PERIFERICI, DI BURKITT	LINFOCITI B MATURI	BURKITT		
3	LINFOMA DI DERIVAZIONE DAI LINFOCITI B PERIFERICI, MANTELLARE	LINFOCITI B MATURI	MANTELLARE	NODALE	CLASSICO

ICD-O | ICC

ICD-11 | WHO

AI MODELS: GENOMIC VARIANTS & ANNOTATIONS

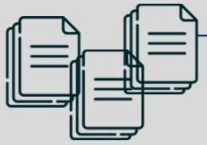
Clinical Data Platform

Pathology Reports

BIOMARKERS

IDistologico IRiferimento	DataPrelievo	DataReport	Diagnosi	TipoTest	Cellularita	TipoMutazione	Gene	Exone	Nomenclatura	Mappatura	VAF	Livello	TipoAlterazione
123	17/04/2024	29/04/2024	Carcinoma endometrioide di alto grado	Oncomine Comprehensive Assay (v3)	60%	SNV	ATRX	33	p.Glu2333Ter	c.6997G>T	25%	III	Patogenica
123	17/04/2024	29/04/2024	Carcinoma endometrioide di alto grado	Oncomine Comprehensive Assay (v3)	60%	SNV	FBXW7	12	p.Arg658Ter	c.1972C>T	34%	III	Patogenica
123	17/04/2024	29/04/2024	Carcinoma endometrioide di alto grado	Oncomine Comprehensive Assay (v3)	60%	SNV	RHOA	2	p.Gly17Glu	c.50G>A	5%	III	Patogenica

VCF



VCF METADATA

vcfID	vcfVersion	vcfTypeID	seqPlatform	seqSoftware	panel	BAMSoftware	fileFormat	fileUTCtime	sampleGender	reference	phasing	TS_TV_Ratio	transitionsCount	transversionsCount
123	v1	I	termofisher	termofisher	Custom	tvc 5.6-11 (85f0861) - Torrent Variant Caller	VCFv4.1	2018-05-08T05:48:18	Unknown	hg19	none	0.0	["AG":0,"CT":0,"GA":0,"TC":0}	{"AC":0,"AT":0,"CA":0,"CG":0,"GC":0,"GT":0,"TA":0,"T
123	v1	I	termofisher	termofisher	Custom	tvc 5.6-11 (85f0861) - Torrent Variant Caller	VCFv4.1	2018-04-28T06:59:48	Unknown	hg19	none	2.0	["AG":1,"CT":1,"GA":0,"TC":0}	{"AC":0,"AT":0,"CA":0,"CG":0,"GC":1,"GT":0,"TA":0,"T
123	v1	I	termofisher	termofisher	Custom	tvc 5.6-11 (85f0861) - Torrent Variant Caller	VCFv4.1	2018-04-28T06:52:04	Unknown	hg19	none	3.0	["AG":2,"CT":0,"GA":1,"TC":0}	{"AC":0,"AT":0,"CA":0,"CG":0,"GC":0,"GT":0,"TA":0,"T
123	v1	I	termofisher	termofisher	Custom	tvc 5.6-11 (85f0861) - Torrent Variant Caller	VCFv4.1	2018-04-28T06:49:03	Unknown	hg19	none	1.0	["AG":1,"CT":0,"GA":1,"TC":0}	{"AC":0,"AT":0,"CA":0,"CG":0,"GC":1,"GT":0,"TA":1,"T
123	v1	I	termofisher	termofisher	Custom	tvc 5.6-11 (85f0861) - Torrent Variant Caller	VCFv4.1	2018-05-08T08:08:19	Unknown	hg19	none	0.33	["AG":1,"CT":0,"GA":1,"TC":0}	{"AC":0,"AT":0,"CA":0,"CG":1,"GC":2,"GT":0,"TA":0,"T

AI MODELS: OUTCOME EVALUATION

Clinical Data Platform

Clinical Variables

Surveys

OUTCOME EVALUATION

Value Based Medicine

AI MODELS: IMAGING DIAGNOSIS

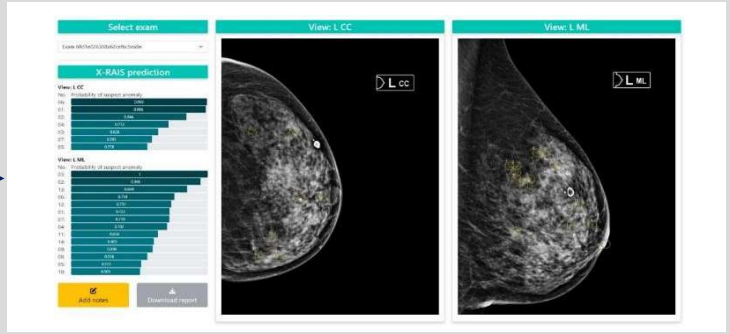
Clinical Data Platform

Clinical Variables

Imaging

IMAGING DIAGNOSES

X-Rais



NEXT STEPS

- Increase technological and scientific management collaboration
- Technological consolidation
- Generate and use synthetic data
- Medical algorithms and tools certification
- Comply with privacy regulations

THANK YOU

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Bigoni Ilaria – LAIFE REPLY