

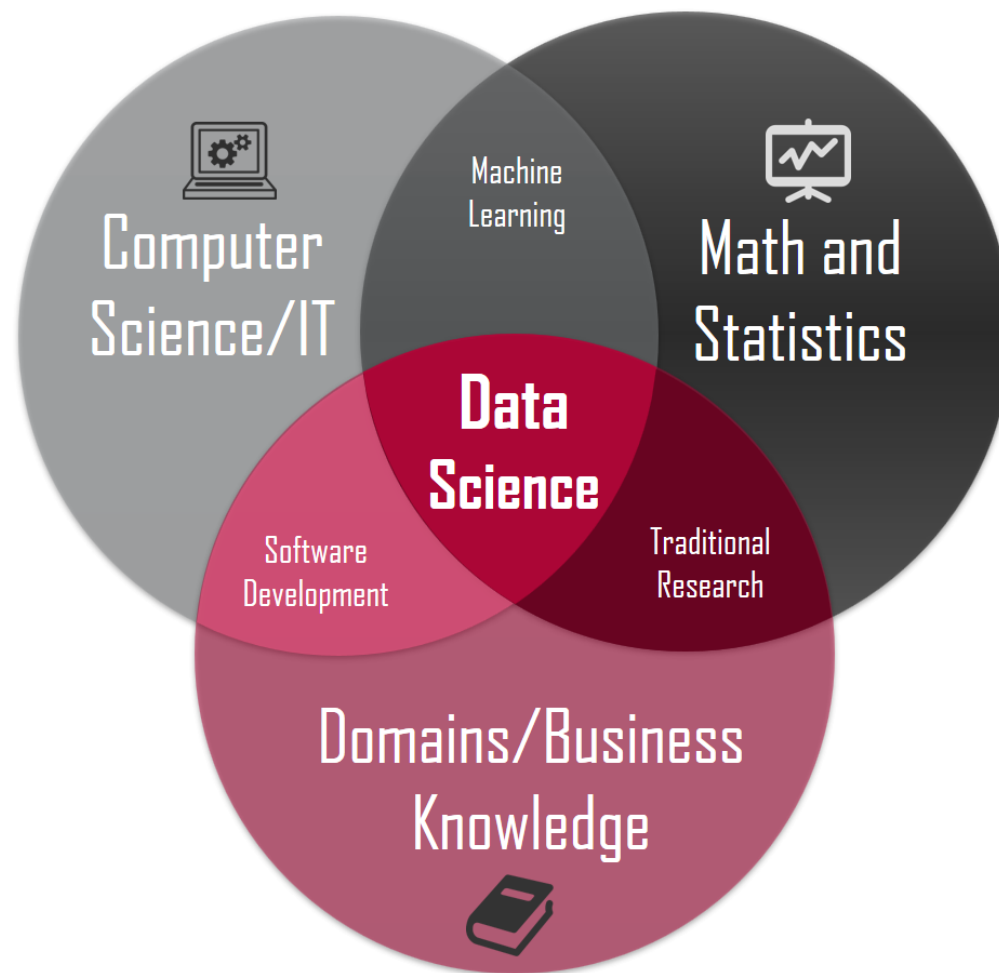
Big Data w analizie danych
klinicznych i genetycznych
na przykładzie
personalizacji leczenia
ostrej białaczki
limfoblastycznej



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Rewolucje technologiczne

- Big Data
- Data Science
- Genetyka



Strategmed III

- Śląski Uniwersytet Medyczny w Katowicach,
- Uniwersytet Medyczny w Łodzi,
- Instytut Genetyki Człowieka Polskiej Akademii Nauk z siedzibą w Poznaniu,
- Uniwersytet Mikołaja Kopernika w Toruniu,
- Collegium Medicum im. Ludwika Rydygiera w Bydgoszczy
- Uniwersytet Medyczny w Lublinie,
- Uniwersytet Medyczny im. Piastów Śląskich we Wrocławiu
- Uniwersytet Medyczny im. Karola Marcinkowskiego w Poznaniu
- Warszawski Uniwersytet Medyczny
- Uniwersytet Medyczny w Białymstoku
- Gdański Uniwersytet Medyczny
- Pomorski Uniwersytet Medyczny w Szczecinie



Narodowe Centrum
Badań i Rozwoju



Wybrane cele projektu

- Wdrożenie nowej procedury leczenia AIEOP-BFM ALL 2017
- Dalsza poprawa skuteczności leczenia
- Udostępnienie platformy naukowej z bazą danych (w tym zebranych podczas realizacji projektu)

Aplikacja PersonALL

Menu > Patients zone

+ Add

Edit

Delete

Medical record

Move Patient's Data

Activate for Marvin

Name	Global code	Local code	Identification no.
	0008	3	
	0009	4	
	0010	5	
	0011	6	
	0012	7	
	0013	8	
	0014	9	

Menu > Patients zone > Medical record > Edit diagnosis

Global code: 0007

Local code: ZA-0007

Basic information

Medical evaluation

Cytometry

Molecular examinations

Cytogenetics

Sequencing

Documents

Microdeletions

Point mutations

Gene fusions

Gene expressions

NGS

Gene rearrangement

Analysis of point mutations

Delete?	Gene	DNA location	Protein location	Ref. sequence
☐	CRLF2	+	+	None
☐	IL7R	+	+	None
☐	JAK1	+	+	None
☐	JAK2	+	+	None
☐	P53	+	+	None

Comment:

Save

Cancel

Global code: 0007

Local code: ZA-0007

Basic information

Medical evaluation

Cytometry

Molecular examinations

Basic information

Date of diagnosis: 2012-05-14

Type of diagnosis: Primary disease

Eligible for study: No

Reason for not eligible for study: Significant previous treatment

Current therapy: AIEOP-BFM ALL 2017

Weight (kg) at diagnosis: 14,60

Height (cm) at diagnosis: 101,50

Body surface (m2) at diagnosis: 0,63

BM at diagnosis: Not performer

%BM blasts at diagnosis: 94,80

BM comment:

Version 1.1.3.1

Treatment details

PCR-MRD at TP1: <=10^-4 (negative)

PCR-MRD at TP2: 0 (negative)

First Risk Group: early non-SR

Wsparcie procedury leczenia

Edit phase - Medical rec: X

Bezpieczna | <https://personall.net/PatientsZone/TherapyPhases/EditTherapyPhase/3?patientId=5>

Menu > Patients zone > Medical record > Edit phase

Logged as: Marek Sikora | Logout

Edit phase

Global code: 0005

Local code: ZA-0005

Age: 13

Gender: Male

Actual RG: HR

Identification no.: not available

Patient: not available

Basic information

Phase name: * Consolidation A

Start date * 2012-03-26

End date: 2012-06-01

Body surface: 0,90

Medication

Additional MTX dose: No

Deviation in timing: No

Deviation in medication: No

Deviation in dosage: No

Deviation cause:

Deviation description:

Specify deviation: ☐

Diagram

Raportowanie i eksport danych

00010/0088

S-ALT/S-AST:	NCI grade I	Creatinine:	NCI grade 0
Proteinuria:	NCI grade 0	Hematuria:	NCI grade 0
Creatinine clearance:	NCI grade 0	Arrythmia:	NCI grade 0
Cardiac function:	NCI grade 0	Echocardio: LV-SF:	NCI grade 0
Central neurotoxicity:	NCI grade 0	Peripheral neurotoxicity:	NCI grade 0
Osteonecrosis:	NCI grade 0	Other toxicity:	No

Phase Protocol IB/1: 28-09-2015 - 31-10-2015

Medication			
Deviation in timing:	Yes	Deviation in medication:	No
Deviation in dosage:	No	Deviation cause:	Other toxicity
Deviation description:	fever		
Toxicities			
General condition:	NCI grade II	Infection:	NCI grade II
Fever:	NCI grade II	Nausea:	NCI grade I
Vomiting:	NCI grade I	Stomatitis:	NCI grade 0
S-bilirubin:	NCI grade 0	Diarrhea:	NCI grade 0
S-ALT/S-AST:	NCI grade I	Creatinine:	NCI grade 0
Proteinuria:	NCI grade 0	Hematuria:	NCI grade 0
Creatinine clearance:	NCI grade 0	Arrythmia:	NCI grade 0
Cardiac function:	NCI grade 0	Echocardio: LV-SF:	NCI grade 0
Central neurotoxicity:	NCI grade 0	Peripheral neurotoxicity:	NCI grade 0
Osteonecrosis:	NCI grade 0	Other toxicity:	No

Health summary

18-12-2017

Name:	Identification no.:
Code: 00010/0088	Date of birth: 31-01-2014
Age: 3	Gender: Female
Country: Poland	Registration date: 01-12-2017
Risk group:	

Diagnosis: 17-08-2015

Basic information			
Type of diagnosis:	Primary disease	Eligible for study:	Yes
Current therapy:	AIEOP-BFM ALL 2017	Weight (kg) at diagnosis:	13,00
Height (cm) at diagnosis:	96,00	Body surface (m2) at diagnosis:	0,59
%BM blasts at diagnosis:	95,20	WBC at diagnosis:	7520
Platelets:	46000,00	%Blasts (PB) at diagnosis:	27
HB (g/dl):	6,10	Hepatomegaly:	Yes
Hepatomegaly cm below costal margin:	1,50	Splenomegaly:	No
Mediastinal mass:	No	Preexisting disease:	No
CSF contaminated with blood:	Yes	Cranial nerve palsy:	No
Cerebral mass:	No	CNS status at diagnosis:	CNS status 2
Medical evaluation			
Immunophenotype:	T-ALL	Immunophenotype details:	Common (cortical) T
Karyotype t(4;11):	Negative	Karyotype t(9;22):	Negative
Other karyotypes 1:	+X	Other karyotypes 2:	random aberration not in this list
Hypodiploidy:	>=50	Hyperdiploidy:	>=50
KMT2A-AFF1(MLL-AF4):	Negative	ETV6-RUNX1 (TEL-AML1):	Positive
BCR/ABL:	Negative	Pre-phase cumulative dose of prednisone (mg/m2):	251
WBC/μL - on day 8:	3620	Blast cell count/μL on day 8:	0
WBC/μL - on day 15:	2770	Blast cell count/μL on day 15:	0
CR1 achieved:	Yes	Timing of CR1:	Early (day 33)
Date of CR1:	25-09-2015	Day of CR1:	39

Markers			
CD10(%):	100,00	CD19(%):	100,00
CD33(%):	0,00	cytCD3(%):	0,00

Phase Protocol IA - Dexa: 17-08-2015 - 27-09-2015

Medication			
Deviation in timing:	Yes	Deviation in medication:	No
Deviation in dosage:	No	Deviation cause:	Other toxicity
Deviation description:	upper respiratory tract infection, diarrhea		
Toxicities			
General condition:	NCI grade II	Infection:	NCI grade II
Fever:	NCI grade II	Nausea:	NCI grade 0
Vomiting:	NCI grade 0	Stomatitis:	NCI grade 0
S-bilirubin:	NCI grade 0	Diarrhea:	NCI grade II

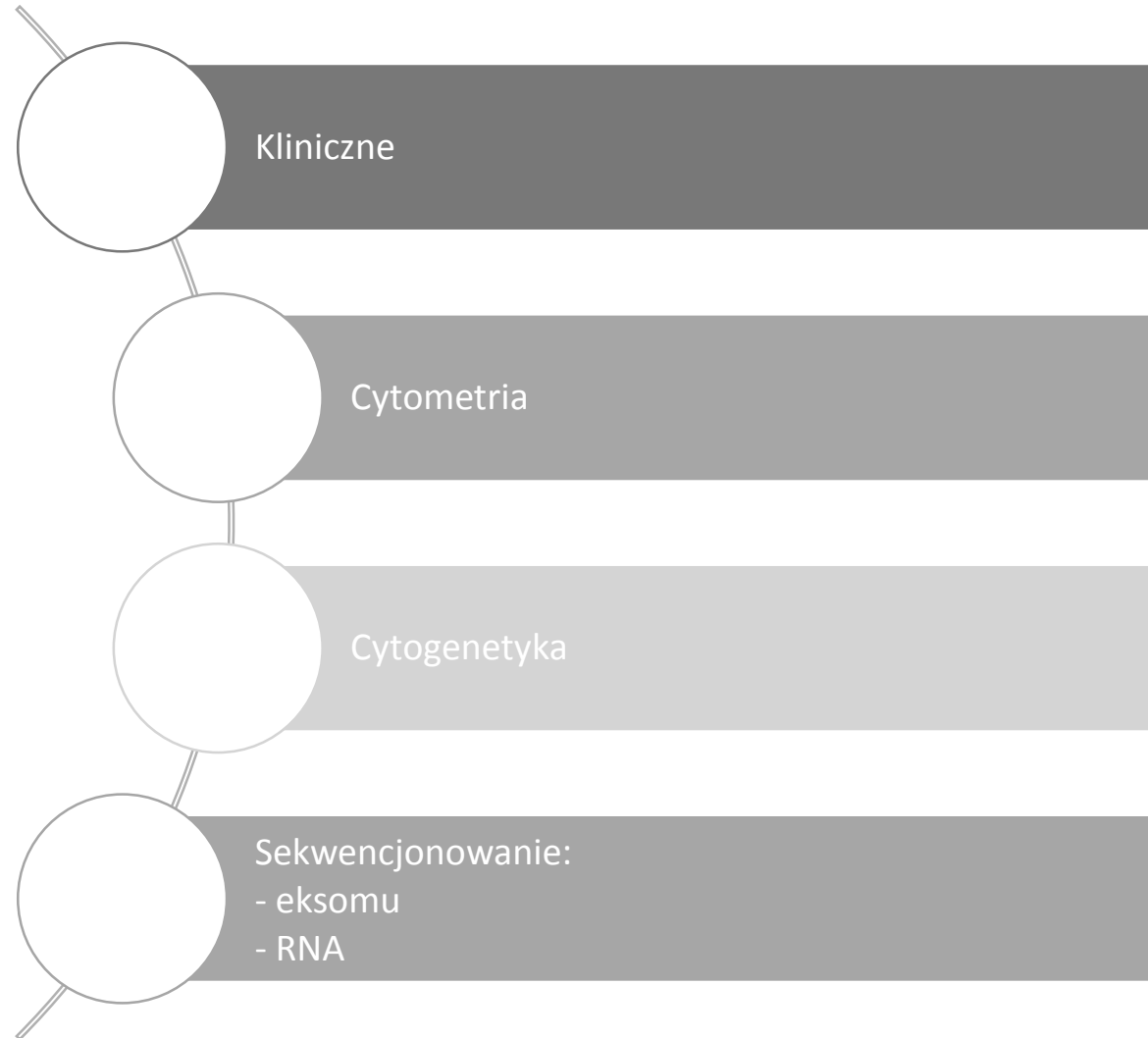
Dane diagnostyczne i genetyczne

Poziomy przechowywania danych

- L1 - dane surowe
- L2 - dane pośrednie
- L3 - dane wpisywane wprost do systemu



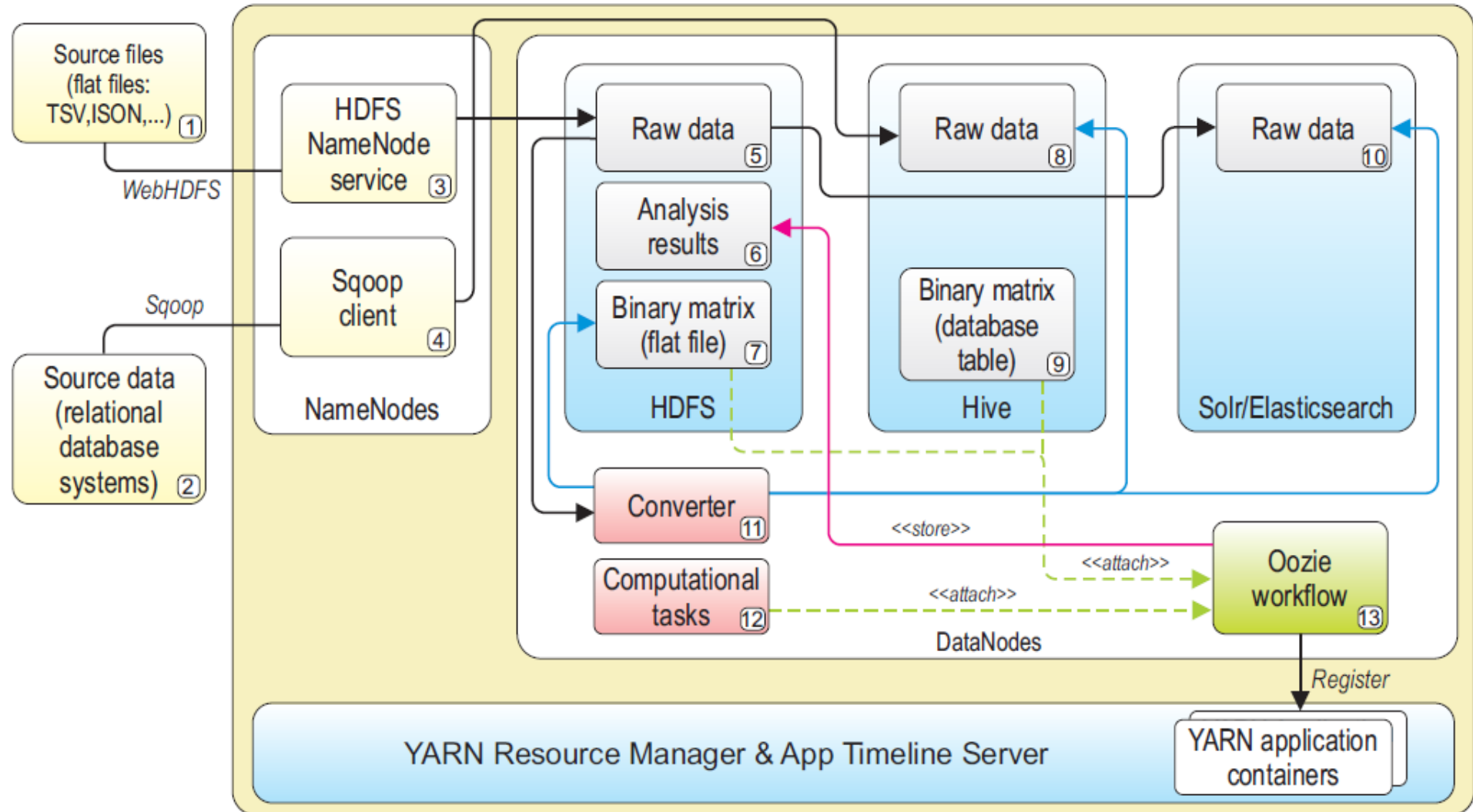
Źródła danych diagnostycznych



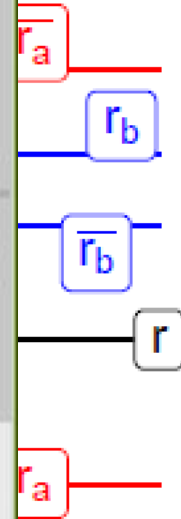
Źródła danych:

- RNA-seq i mikromacierze oligonukleotydowe - badanie zmian w profilu ekspresji genów
- WES/WGS - identyfikacja mutacji somatycznych oraz indeli (insercji/delecji)
- Mikromacierze SNP - identyfikacja zmian liczby kopii genów (CNV)
- DMH (Differential Methylation Hybridization) - zmiany w profilu metylacji wysp CpG
- miRNA-seq - zmiany profilu ekspresji miRNA

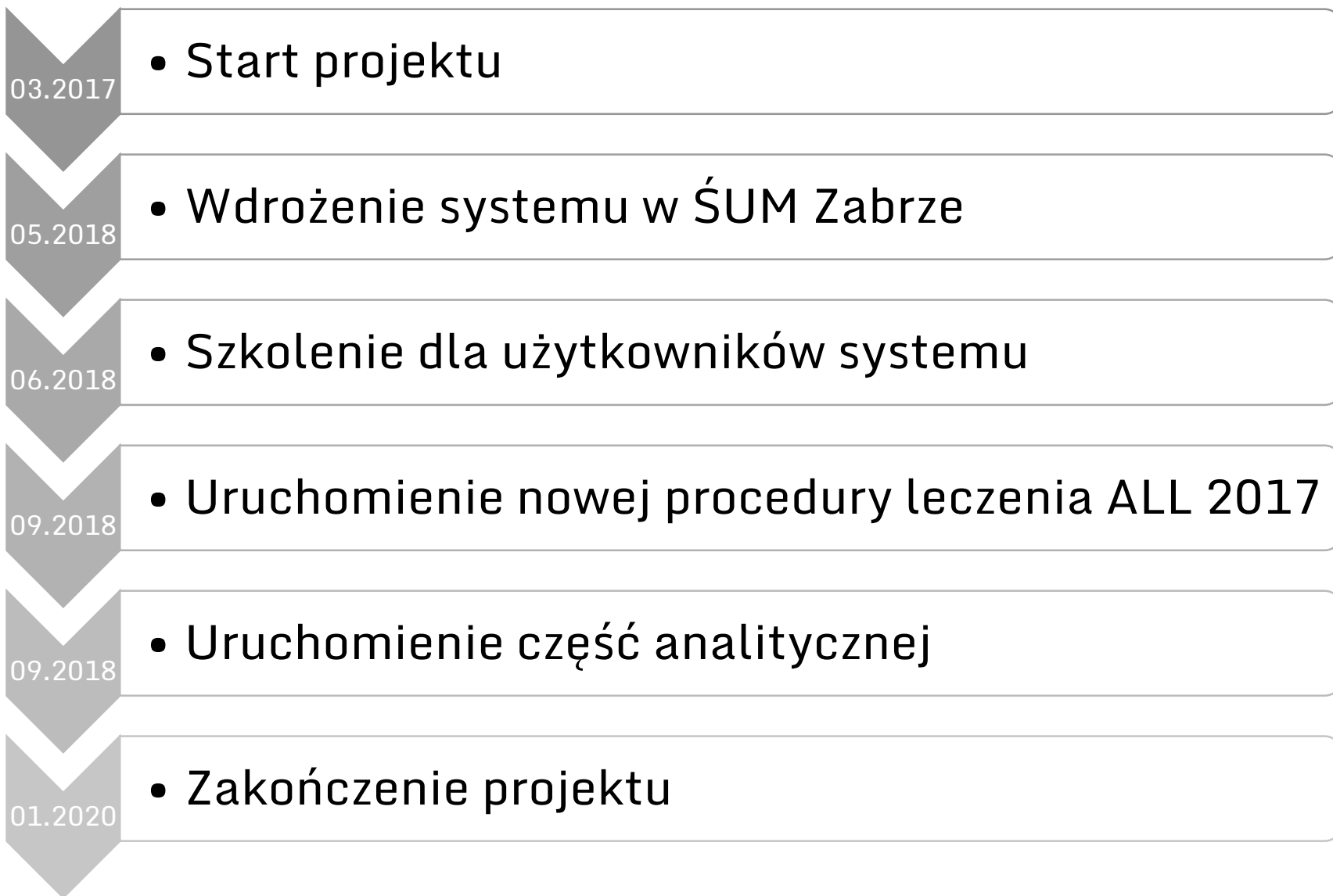
Architektura



- **F** p
z
- **P** a
- **A**



Stan realizacji projektu



SILMINE STWORZONE PRZEZ: **3SOFT**  netology

SILMINE

WE KNOW
YOU KNOW.

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