



Bulut Tabanlı Sistemlerde Genetik Araştırmalar: Bioconductor Deneyimi

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Microsoft Research

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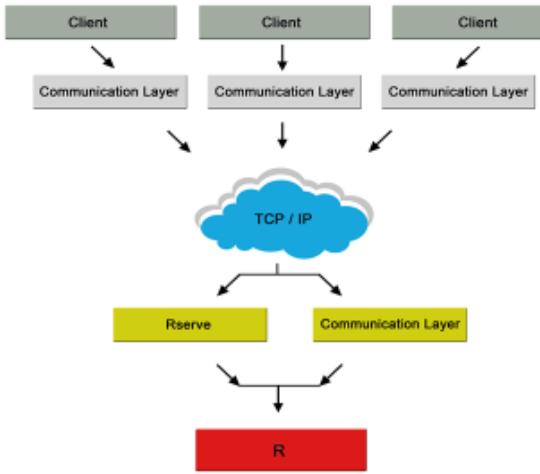


[Hacettepe Üniversitesi Biyoistatistik Ana Bilim Dalı](#)

Tez Bilgileri Detay

Tez No İndirme	Tez Künye	Durumu
358990	New approach to unsupervised based classification on microarray data / Mikrodizilim verilerden danışmansız öğrenmeye dayalı sınıflama yeni yaklaşım Yazar: ERDAL COŞGUN Danışman: PROF. DR. ERGUN KARAĞAOĞLU Yer Bilgisi: Hacettepe Üniversitesi / Sağlık Bilimleri Enstitüsü / Biyoistatistik Ana Bilim Dalı / Biyoistatistik Bilim Dalı Konu: Biyoistatistik = Biostatistics Dizin: Bilgisayar yazılımları = Computer softwares ; Gen ifadesi = Gene expression ; Rastgele modeller = Random models ; Veri madenciliği = Data mining ; İstatistiksel modeller = Statistical models	Onaylandı Doktora İngilizce 2013 109 s.

Client Structure of Gene3E



Rgenetik

2009- 2012

DAVID Bioinformatics Database

Functional Annotation Database

Home Start Analysis Shortcut to DAVID Tools Technical Center Downloads 8

*** Announcing the new DAVID Web Service which allows access to DAVID

Upload List Background

Gene List Manager

Select to limit annotations by one or more species [Help](#)

- Use All Species - Homo sapiens(1)
- Disease (1 selected)
- Functional_Categories (2 selected)
- Gene_Ontology (3 selected)
- General Annotations (0 selected)
- Literature (0 selected)
- Main_Accessions (0 selected)
- Pathways (1 selected)
- Protein_Domains (2 selected)
- Protein_Interactions (0 selected)
- Tissue_Expression (0 selected)

Annotation Summary Result

Current Gene List: List_1

Current Background: Homo sapiens

List Manager Help

List_1

Select List to:

- Use Rename
- Remove Combine
- Show Gene List

Combined View for Selected Annotation

Functional Annotation Clustering

Random Forest Trial 100 Finished.

Progress

RUN!

Genotype File: C:\Users\Ali\workspace\RWrapper2\data\colon.csv

Label File: C:\Users\Ali\workspace\RWrapper2\data\class.csv

Methods

Dimension Reduction Method: Independent Component Analysis

Generalization Method: Bootstrap

Clustering: --None--

Classification: Random Forest



Genetik Çalışmalar



Precision medicine 😊



Erken Tanı



Hastalık Risk
Tahminleme



Kişileştirilmiş
Tıp



Hedef
İlaç



Gen
Terapisi

Araştırma Enstitüleri

Akademik Tıp
Merkezleri

İlaç Şirketleri

Sağlık Sistem
Sağlayıcıları

Klinik Araştırma

İlaç geliştirme

Klinik Karar Destek
Sistemleri

Açık Kaynak Genetik Projeler ve R Uygulamaları



Araştırma

Genomics Notebooks

Azure'da Bioconductor

Genetik Veri Bilimi VM

Genetik Veri Tabanları



Otomasyon - Ölçeklendirme

Cromwell on Azure
workflow & task execution engine



Genetik verinin geleceği?



Yeni Nesil Sekanslama...

İlaç tasarımı

Klinik Uygulamalar



Sekanslanan genom sayısı

240K

2014

1.6M

2017

~5M

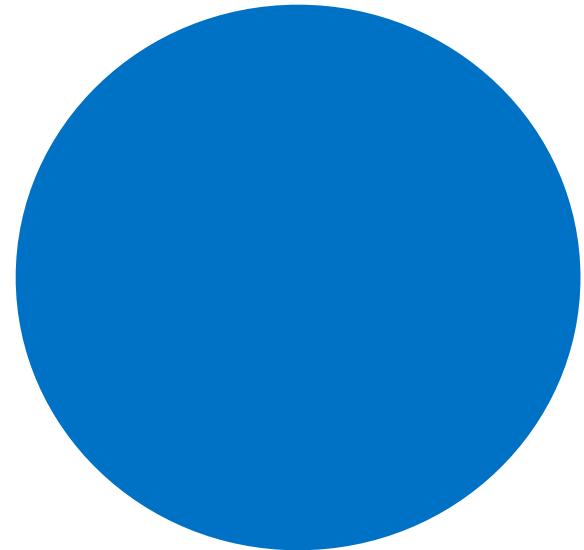
2020

100M

2030

Tahmini

Veri Büyüklüğü



Whole genome

100_{GB}
\$1000



Whole **exome**

5_{GB}
\$200



Genotype

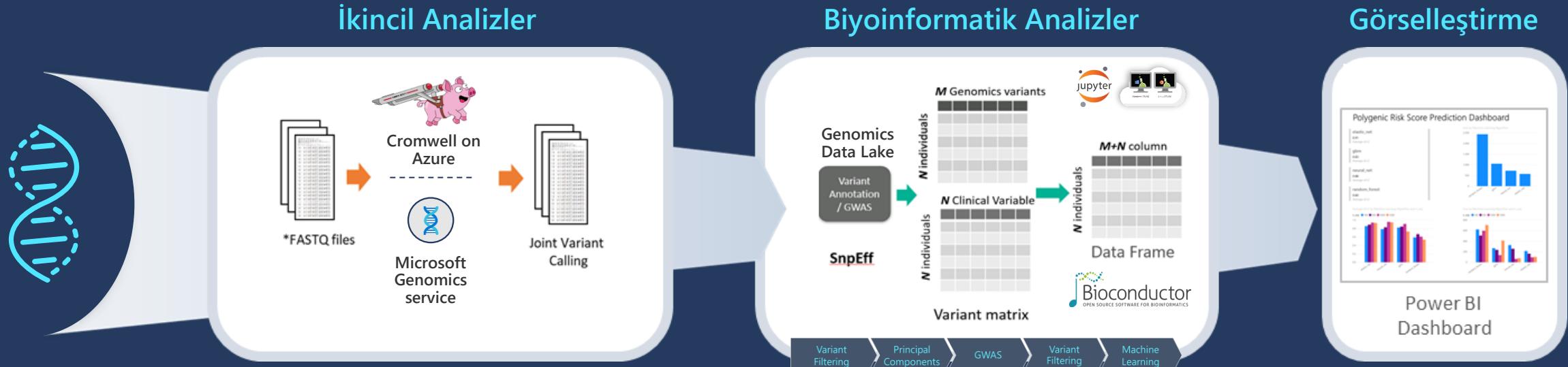
1_{MB}
\$50



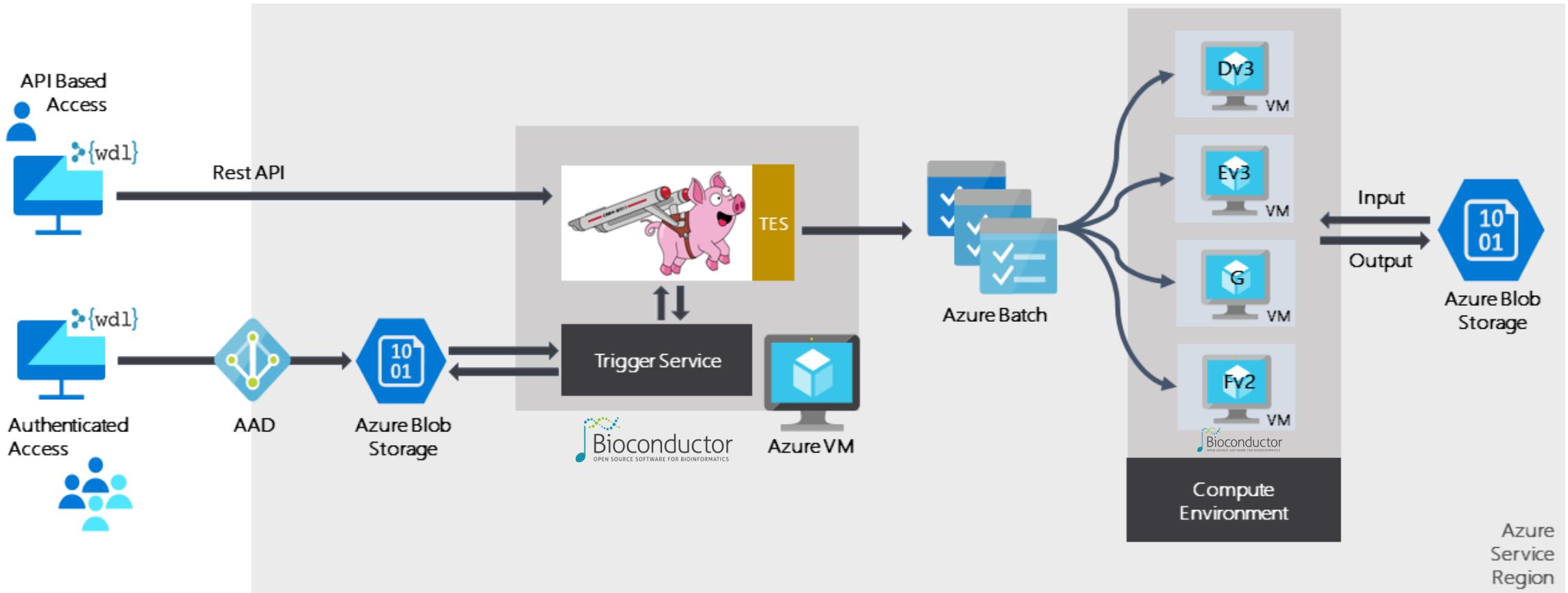
Gene panel

1_{KB}
\$20

E2E- Genetik Data Science Şablonları



R Otomasyonu - Cromwell Örneği



<https://github.com/microsoft/CromwellOnAzure>



About Bioconductor

Bioconductor provides tools for the analysis and comprehension of high-throughput genomic data.

Bioconductor uses the R statistical programming language, and is open source and open development. It has two releases each year, and an active user community. Bioconductor is also available as an [AMI](#) (Amazon Machine Image) and [Docker](#) images.

News

- Bioconductor [3.12](#) is available.
- See our [google calendar](#) for events, conferences, meetings, forums, etc. Add your event with email to events at bioconductor.org.
- Core team **job opportunities** available, contact Martin.Morgan at RoswellPark.org
- Bioconductor [F1000 Research Channel](#) is available.
- Orchestrating single-cell analysis with Bioconductor ([abstract](#); [website](#)) and other [recent literature](#).

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Search:

BioC 2021

Visit the [BioC 2021](#) website for complete conference information! The virtual conference will be held August 4-6, 2021!

News highlights:

- Abstract Submission Deadline for talk, demo, long workshop, digital poster, or Birds-of-a-feather is next Tuesday March 16! Submit [here](#)
- Registration opening soon.
- See the list of confirmed speakers on the [website home page](#)

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Project Leads:
Martin Morgan
Nitesh Turaga
Erdal Cosgun

[Bioconductor - Docker for Bioconductor](#)



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Docker containers for Bioconductor

Docker packages software into self-contained environments, called containers, that include necessary dependencies to run. Containers can run on any operating system including Windows and Mac (using modern Linux kernels) via the [Docker engine](#).

Containers can also be deployed in the cloud using [Amazon Elastic Container Service](#), [Google Kubernetes Engine](#) or [Microsoft Azure Container Instances](#)

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- [Modifying Image Container](#)
- [Singularity](#)
- [Microsoft Azure Container Instances](#)
 - [Using containers hosted on Microsoft Container Registry](#)
 - [Use Azure Container Instances to run bioconductor images on-demand on Azure](#)
- [How to contribute](#)
- [Acknowledgements](#)

MCR- Bioconductor.org

<https://bioconductor.org/help/docker/#msft>

- Entegre Azure File Share
- Azure CLI

Microsoft Azure Container Instances

If you are a Microsoft Azure user, you have an option to run your containers using images hosted on [Microsoft Container Registry](#).

Microsoft Container Registry (MCR) is the primary Registry for all Microsoft Published docker images that offers a reliable and trustworthy delivery of container images with a syndicated catalog

Using containers hosted on Microsoft Container Registry

You can learn more about the `bioconductor_docker` image hosted on Microsoft Container Registry [here](#).

Pull the `bioconductor_docker` image from Microsoft Container Registry, specifying your `tag` of choice. Check [here](#) for the list of tags under "Full Tag Listing":

```
docker pull mcr.microsoft.com/bioconductor/bioconductor_docker:<tag>
```

To pull the latest image:

```
docker pull mcr.microsoft.com/bioconductor/bioconductor_docker:latest
```

Example: Run RStudio interactively from your docker container

To run RStudio in a web browser session, run the following and access it from `127.0.0.1:8787`. The default user name is "rstudio" and you can specify your password as the example below (here, it is set to 'bioc'):

```
docker run --name bioconductor_docker_rstudio \
-v ~/host-site-library:/usr/local/lib/R/host-site-library \
-e PASSWORD='bioc' \
-p 8787:8787 \
mcr.microsoft.com/bioconductor/bioconductor_docker:latest
```

To run RStudio on your terminal:

```
docker run --name bioconductor_docker_rstudio \
-it \
-v ~/host-site-library:/usr/local/lib/R/host-site-library \
-e PASSWORD='bioc' \
-p 8787:8787 \
mcr.microsoft.com/bioconductor/bioconductor_docker:latest R
```

Bioconductor için Docker Hub UI

https://hub.docker.com/_/microsoft-bioconductor-bioconductor-docker

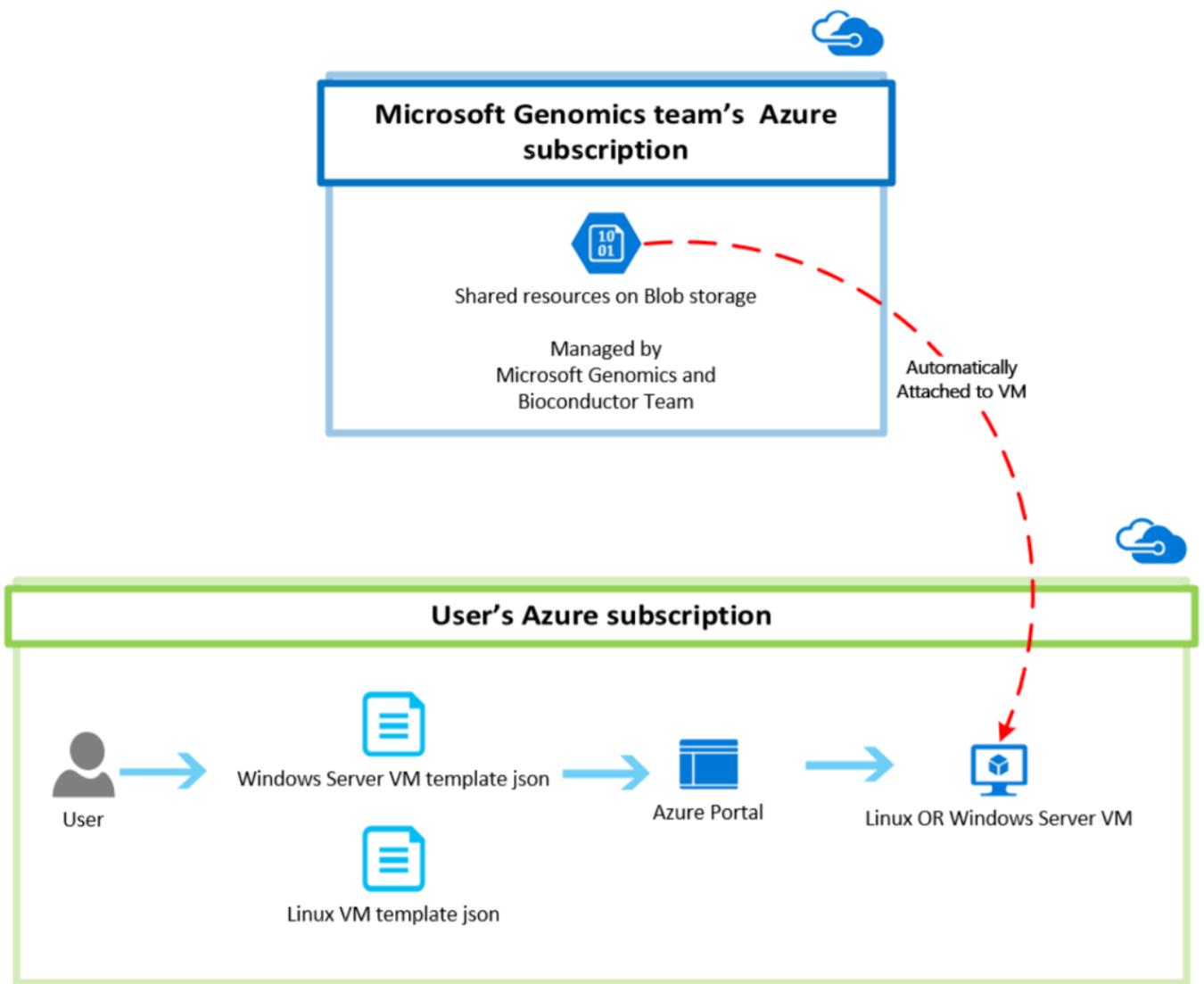
The screenshot shows the Docker Hub page for the 'microsoft/bioconductor_docker' image. At the top, there's a navigation bar with 'Explore' and 'Official Bioconductor image mirror'. Below the navigation is a large image of the Windows logo (four colored squares). To the right of the image is the title 'Official Bioconductor image mirror' and the author 'By Microsoft'. A download count of '5.2K' is displayed below the image. Below the image are three tabs: 'Container', 'x86-64', and 'Base Images'. The 'Container' tab is selected. The main content area contains sections for 'Using Bioconductor images on Azure', 'About bioconductor', 'Related Repos', 'How to Use this Image', and 'Support and feedback'. The 'Using Bioconductor images on Azure' section includes a code snippet for pulling the image and instructions for running RStudio interactively.

The screenshot shows the Docker Hub page for the 'microsoft/bioconductor_docker' image. At the top, there's a navigation bar with 'Explore' and 'Official Bioconductor image mirror'. Below the navigation is a large image of the Windows logo (four colored squares). To the right of the image is the title 'Official Bioconductor image mirror' and the author 'By Microsoft'. A download count of '5.2K' is displayed below the image. Below the image are three tabs: 'Container', 'x86-64', and 'Base Images'. The 'Container' tab is selected. The main content area contains sections for 'Description', 'Reviews', and 'Resources'. The 'Description' section contains a large heading 'Using Bioconductor images on Azure' and a 'Featured Tags' section with a single item: 'latest docker pull mcr.microsoft.com/bioconductor/bioconductor_docker:latest'.

Featured Tags

- latest `docker pull mcr.microsoft.com/bioconductor/bioconductor_docker:latest`

Azure R-Bioconductor Data Science VM



VM Şablonları

Windows and Linux VM Paketleri

1. Bioconductor Genomics Data Science Virtual Machine - Windows Server 2016

The 'Data Science Virtual Machine (DSVM)' is a 'Windows Server 2019 with Containers' VM & includes popular tools for data exploration, analysis, modeling & development.

Highlights:

- Anaconda Python
- SQL Server 2019 Dev. Edition - With In-Database R and Python analytics
- Microsoft Office 365 ProPlus BYOL - Shared Computer Activation
- Julia
- Jupyter notebooks
- Visual Studio Community Ed. + Python, R & node.js tools
- Power BI Desktop
- Deep learning tools e.g. TensorFlow, Chainer
- ML algorithm libraries e.g. xgboost, Vowpal Wabbit
- Microsoft Teams
- Azure SDKs + libraries for various Azure Cloud offerings. Integration tools are included for:

1. Azure Machine Learning
2. Azure Data Factory
3. Stream Analytics
4. SQL Data Warehouse
5. Hadoop + Apache Spark (HDInsight)
6. Data Lake
7. Blob storage
8. ML & Data Science tutorials as Jupyter notebooks
9. R-Bioconductor workflows and samples
10. Microsoft Genomics notebooks
11. GATK 4.1.8

This image is pre-configured with Nvidia drivers, CUDA Toolkit, & cuDNN library for GPU workloads available if using [NC class VM SKUs](#).

2. Bioconductor Genomics Data Science Virtual Machine - Ubuntu 18.04

The **Data Science Virtual Machine - Ubuntu 18.04** (DSVM) is an Ubuntu-based virtual machine image that makes it easy to get started with machine learning, including deep learning, on Azure.

Highlights:

1. Anaconda Python
2. Jupyter, JupyterLab, and JupyterHub
3. Deep learning with TensorFlow and PyTorch
4. Machine learning with xgboost, Vowpal Wabbit, and LightGBM
5. Julia
6. Azure SDKs and libraries
7. Azure Machine Learning SDKs and sample notebooks
8. R support
9. Spark
10. R-Bioconductor workflows and samples
11. Microsoft Genomics notebooks
12. GATK 4.1.8

This image is pre-configured with NVIDIA drivers, CUDA Toolkit, and cuDNN library for GPU workloads if using [NC class VM SKUs](#).

Size	vCPU	Memory: GiB	Temp storage (SSD) GiB	GPU	GPU memory: GiB	Max data disks	Max NICs / Expected network bandwidth (Mbps)
Standard_NC4as_T4_v3	4	28	180	1	16	8	2 / 8000
Standard_NC8as_T4_v3	8	56	360	1	16	16	4 / 8000
Standard_NC16as_T4_v3	16	110	360	1	16	32	8 / 8000
Standard_NC64as_T4_v3	64	440	2880	4	64	32	8 / 32000

main · 1 branch · 0 tags

[Go to file](#) [Code](#)

erdalcosgun	Update Azure Storage Explorer doc v.0.2	fb16a3c yesterday	43 commits
docs	Update Azure Storage Explorer doc v.0.2	yesterday	
genomics-data-science-vm	Update genomics_vm_template_linux.json	2 months ago	
sample-notebooks	GATK bundle and ENCODE sample notebooks	12 days ago	
CODE_OF_CONDUCT.md	Initial CODE_OF_CONDUCT.md commit	9 months ago	
LICENSE	Initial LICENSE commit	9 months ago	
README.md	add GATK resource bundle & ENCODE sample notebooks	12 days ago	
SECURITY.md	Initial SECURITY.md commit	9 months ago	

README.md

Genomics Data Analysis with Jupyter Notebooks on Azure

Jupyter notebooks are a great tool for data scientists who are working on genomics data analysis. In this repo, we demonstrate the use of [Azure Notebooks](#) for genomics data analysis via GATK, Picard, Bioconductor and Python libraries.

Here is the list of sample notebooks on this repo:

- [genomics.ipynb](#) : Analysis from 'uBAM' to 'structured data table' analysis.
- [genomicsML.ipynb](#) : Train Machine Learning models with Genomics + Clinical Data
- [genomics-platinum-genomes.ipynb](#) : Accessing Illumina Platinum Genomes data from [Azure Open Datasets*](#) and to make initial data analysis.
- [genomics-reference-genomes.ipynb](#) : Accessing reference genomes from [Azure Open Datasets*](#)
- [genomics-clinvar.ipynb](#) : Accessing ClinVar data from [Azure Open Datasets*](#)
- [genomics-giab.ipynb](#) : Accessing Genome in a Bottle data from [Azure Open Datasets*](#)
- [SnpEff.ipynb](#) : Accessing SnpEff databases from [Azure Open Datasets*](#)
- [1000 Genomes.ipynb](#) : Accessing 1000 Genomes dataset from [Azure Open Datasets*](#)
- [GATKResourceBundle.ipynb](#) : Accessing GATK resource bundle from [Azure Open Datasets*](#)
- [ENCODE.ipynb](#) : Accessing ENCODE dataset from [Azure Open Datasets*](#)
- [genomics-OpenCRAVAT.ipynb](#) : Accessing OpenCRAVAT dataset from [Azure Open Datasets*](#)
- [Bioconductor.ipynb](#) : Pulling Bioconductor Docker image from [Microsoft Container Registry](#)
- [simtable.ipynb](#) : Simulate NGS data, use Cromwell on Azure OR Microsoft Genomics service for secondary analysis and convert the gVCF data to a structured data table.

About

Jupyter Notebooks on Azure for Genomics Data Analysis

[Readme](#) [MIT License](#)

Releases

No releases published

Packages

No packages published

Languages

- Jupyter Notebook 100.0%

main · [genomicsnotebook / sample-notebooks / simtable.ipynb](#) · Go to file · ...

erdalcosgun add simtable notebook · Latest commit c014cfb on Jan 8 · History

1 contributor

366 lines (366 sloc) 15.7 KB

[Raw](#) [Blame](#) [Edit](#)

Genomics Data Simulation to Machine Learning ready table

Jupyter notebook is a great tool for data scientists who are working on Genomics data analysis. We will demonstrate the process of simulation of paired-end fastq files to downstream analysis ready table format with ART, Cromwell on Azure, GATK and Picard on Jupyter notebook.

Here is the coverage of this notebook:

- Simulate Next Generation Sequencing Data with ART
- Convert fastq paired-end data to uBAM with Cromwell on Azure
- uBAM to VCF with Cromwell on Azure
 - Alignment and Variant Calling with Microsoft Genomics service
- Convert the gVCF file to a table format

Dependencies:

This notebook requires the following libraries:

- Azure CLI
- AzCopy: Please install latest release of the AzCopy: <https://docs.microsoft.com/en-us/azure/storage/common/storage-use-azcopy-v10>
- Cromwell on Azure: Please download the latest release of CoA from: <https://github.com/microsoft/CromwellOnAzure/releases>
- ART: ART is a set of simulation tools to generate synthetic next-generation sequencing reads. Please download the latest version of this tool from: <https://www.niehs.nih.gov/research/resources/software/biostatistics/art/index.cfm>
- Picard: Please download the latest release of the tool from <https://broadinstitute.github.io/picard/>
- Genome Analysis Toolkit (GATK) (*Users need to download GATK from Broad Institute's webpage into the same compute environment with this notebook: <https://github.com/broadinstitute/gatk/releases>*)
- Users need reference genome for using this notebook on their environment: [hg19.fasta](#)

Important information: This notebook is using Python 3.6 kernel

1. Simulate Next Generation Sequencing Data with ART - Sample Code

We recommend to use ART ([quote from the ART's website](#): "ART is a set of simulation tools to generate synthetic next-generation sequencing reads. ART simulates sequencing reads by mimicking real sequencing process with empirical error models or quality profiles summarized from large recalibrated sequencing data") for NGS data simulation.

This is a great tool to simulate a NGS data for different sequencing platforms. Simulated data sets are very close to the real genomics datasets. Users can test their own downstream analysis with the simulated data sets.

In this notebook, we will demonstrate the 'paired sample fastq' simulation with sample codes. Please visit tool's website for further sample codes.

Please download the ART binary files from this [link](#) than just call the code in below.

Based on the information on the manual of ART, parameters of the simulation are defined as follows:

```
-ss --seqSys The name of Illumina sequencing system of the built-in profile used for simulation
```

SnpEff-Genetik Anotasyon ve sonrasında Bioconductor Paketleri

SnpEff & SnpSift

Genomic variant annotations and functional effect prediction toolbox.

[Download SnpEff](#)

Latest version 5.0e (2021-03-09)
Requires Java 12

 Microsoft
SnpEff and SnpSift are hosted by
[Microsoft Genomics](#)

main genomicsnotebook / sample-notebooks / SnpEff.ipynb Go to file ...

erdalcosgun add SnpEff notebook Latest commit 5bab54a on Nov 13, 2020 History

1 contributor

111 lines (111 sloc) | 3.13 KB

Genomics Data Analysis with Azure Jupyter Notebooks- SnpEff

Jupyter notebooks are a great tool for data scientists who is working on Genomics data analysis. We will demonstrate how to download specific SnpEff dataset/database from Azure Open Datasets.

Here is the coverage of this notebook:

1. Download specific database from SnpEff datasets

Dependencies:

This notebook requires the following libraries:

- Azure storage pip install azure-storage-blob==2.1.0. Please visit [this page](#) for frequently encountered problem for this SDK.

Important information: This notebook is using Python 3.6 kernel

1. Getting the SnpEff data from Azure Open Datasets

Several public genomics data has been uploaded as an Azure Open Dataset [here](#). We create a blob service linked to this open datasets. You can find examples of data calling procedure from Azure Open Dataset for SnpEff datasets in below:

1.a.Install Azure Blob Storage SDK

```
In [ ]: pip install azure-storage-blob==2.1.0
```

1.b.Download the targeted file

```
In [ ]: import os
import uuid
import sys
from azure.storage.blob import BlockBlobService, PublicAccess

blob_service_client = BlockBlobService(account_name='datasetsnpeff', sas_token='sv=2019-10-10&st=2020-09-01T00%3A00%3A00Z&se=2050-09-01T00%3A00%3A00Z&si=prod&sr=&sig=iisafo9tGnYBAvxFUMDGMTbsG2z%2FShaihp7jE5dhW%3D')
blob_service_client.get_blob_to_path('dataset/v5_0/hg19', 'NR_039688.1.txt', './NR_039688.1.txt')
```

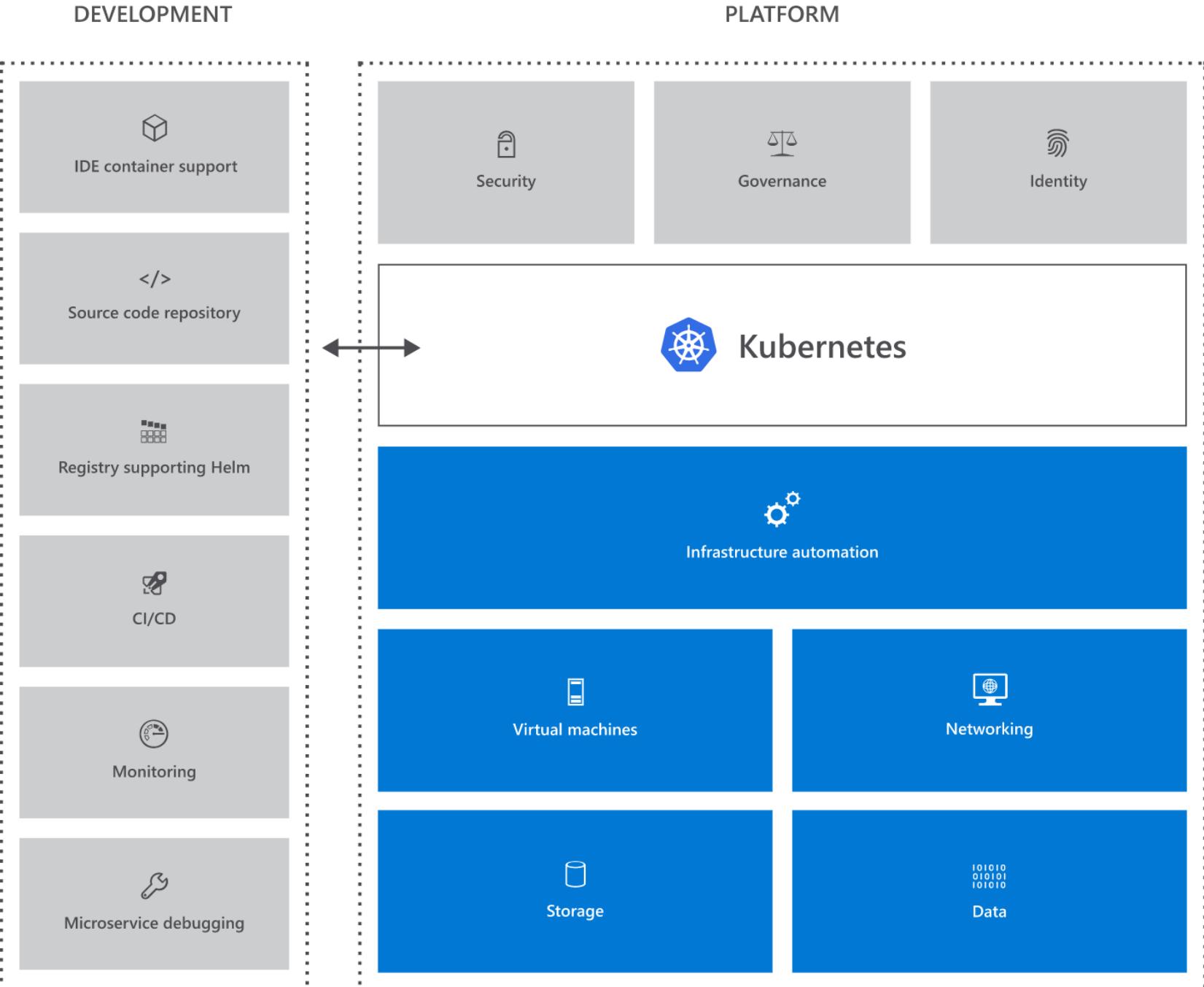
References

1. SnpEff: <https://pcingola.github.io/SnpEff/>

Kubernetes

R Otomasyon

10x Speed



Sudden infant death syndrome (SIDS)

Published Manuscripts

Results from our ongoing studies are published in prestigious peer-reviewed medical journals. Whenever possible these articles have been made open access for parents and clinicians so that they can learn and reference the content.

Five complimentary approaches are at the center of our comprehensive research plan to unravel the causes of SIDS and SUID.



Research pillars:

1. Epidemiology
2. Genetics
3. Physiology
4. Pathology
5. Parental education

Together they form a solid science-based foundation that guides our strategies to prevent further deaths.

A selection of our recent papers are listed below, and organized by these categories.

[Our Research Partners – SIDS Research Guild \(givetostopsids.org\)](#)

Research Studies

AMSRG works with medical researchers and data scientists to study the cause of SIDS, and how it can be prevented in the future. Read more about our current studies and if you can help below.



SUID Research Study

If you are a parent or legal guardian of an infant or child who died suddenly with no apparent medical reason and would like to learn more about this study or how you could participate, email us. We are deeply sorry for your loss.

[Email Us →](#)

A Genetic Database

Researchers at Seattle Children's Research Institute are creating a genetic database to search for key genes or mutations that could explain why some infants/children pass away from SIDS (sudden infant death syndrome), SUID (sudden unexpected infant death), or SUDC (sudden unexplained death in childhood). The research team is led by Dr. Nino Ramirez, Director of the Center for Integrated Brain Research, and Dr. Ghayda Mirzaa, Assistant Professor of Genetic Medicine.

If you joined this study, we would request tissue and/or blood samples and medical records from the deceased child on your behalf as well as saliva cheek swabs from biological family members, such as the child's mother and father. We would then perform genetic testing on these samples and store coded genetic and medical information in a secure database.

Genetik Veri Deposu – Açık Kaynak

- Toplam 3+ petabyte veri

Genetik Veri Tabanları

- Human Reference Genomes
- Illumina Platinum Genomes
- ClinVar variant annotation database
- Genome in a Bottle
- SnpEff variant annotation-prediction database
- gnomAD
- GATK resource bundle
- ENCODE
- OpenCRAVAT

Azure Open Datasets

Easily access curated datasets and accelerate **machine learning**



Quickly build more accurate models



Promote community collaboration



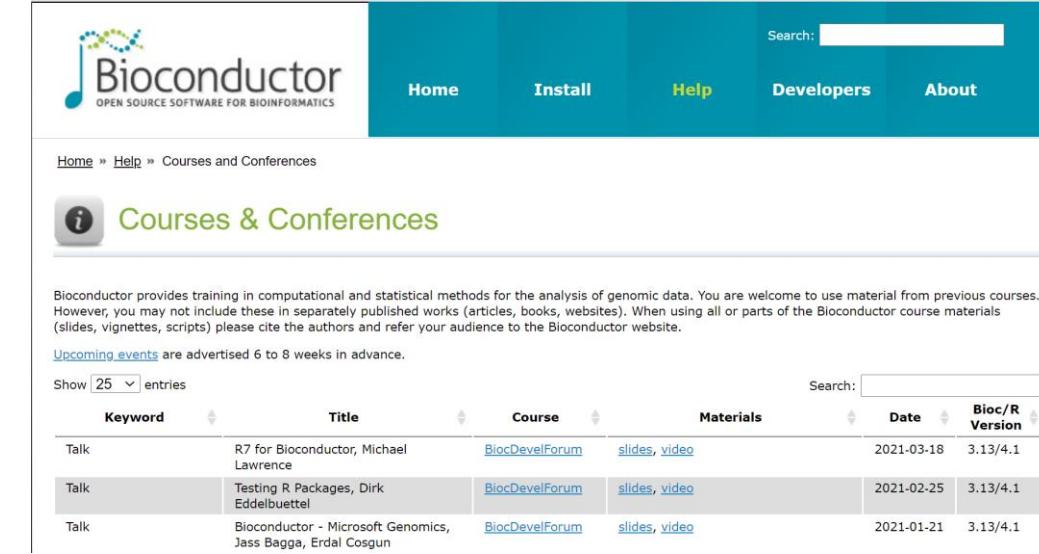
Speed insights with Azure scale



<https://azure.microsoft.com/en-us/services/open-datasets/catalog/genomics-data-lake/>

Kaynaklar

1. [Developers Forum 18 - YouTube](#)-
2. [GitHub - microsoft/genomicsnotebook: Jupyter Notebooks on Azure for Genomics Data Analysis](#)
3. [genomicsnotebook/genomics-data-science-vm at main · microsoft/genomicsnotebook · GitHub](#)
4. [bioconductor \(docker.com\)](#)
5. [SnpEff and SnpSift \(pcingola.github.io\)](#)
6. [Genomics Data Lake - Azure Open Datasets Catalog \(microsoft.com\)](#)



The screenshot shows the Bioconductor website's "Courses & Conferences" section. At the top, there is a navigation bar with links for Home, Install, Help, Developers, and About. A search bar is located in the top right corner. Below the navigation, a breadcrumb trail shows "Home » Help » Courses and Conferences". A title "Courses & Conferences" is displayed with a small info icon. A descriptive text block explains that Bioconductor provides training in computational and statistical methods for the analysis of genomic data, and encourages citation of course materials. It notes that upcoming events are advertised 6 to 8 weeks in advance. A table lists three entries:

Keyword	Title	Course	Materials	Date	Bio/R Version
Talk	R7 for Bioconductor, Michael Lawrence	BioDevelForum	slides , video	2021-03-18	3.13/4.1
Talk	Testing R Packages, Dirk Eddelbuettel	BioDevelForum	slides , video	2021-02-25	3.13/4.1
Talk	Bioconductor - Microsoft Genomics, Jass Bagga, Erdal Cosgun	BioDevelForum	slides , video	2021-01-21	3.13/4.1

II. ULUSLARARASI SAĞLIKTA YAPAY ZEKA KONGRESİ 2021

“COVID-19 PANDEMİSİ”



Uluslararası Konuşmacılar



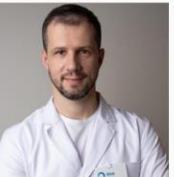
Abdulaziz Ahmed
ABD



Antony Chang
ABD



Erdal Coşgun
ABD



Vasiliy Grachev
RUSYA



Ramesh S. Krishnamurthy
İSVİÇRE



Ilkka Kunnamo
FINLANDIYA



Dana Mukhamejanova
KAZAKSTAN



Salih Tutun
ABD



Eugene Shumilov
ABD



Wija Oortwijn
HOLLANDA



Aydogan Ozcan
ABD



Kristen Yeom
ABD



Bernard P. Zeigler
ABD

Q&A

Teşekkürler

Erdal Cosgun at Microsoft Research

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