

**MOU No.1**

**Memorandum of understanding between CSIR- Institute of Genomics and Integrative Biology CSIR, Delhi and Shri Mathuradas Mohota College of Science, Nagpur**

**Activity/Outcome 1 of MOU No.1**

03 papers published in this work.

The author from the college is Dr. Nikhil V. Palande, Assistant Professor in Zoology.

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## IndiGenomes: a comprehensive resource of genetic variants from over 1000 Indian genomes

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### ABSTRACT

With the advent of next-generation sequencing, large-scale initiatives for mining whole genomes and exomes have been employed to better understand global or population-level genetic architecture. India encompasses more than 17% of the world population with extensive genetic diversity, but is under-represented in the global sequencing datasets. This

gave us the impetus to perform and analyze the whole genome sequencing of 1029 healthy Indian individuals under the pilot phase of the 'IndiGen' program. We generated a compendium of 55,898,122 single allelic genetic variants from geographically distinct Indian genomes and calculated the allele frequency, allele count, allele number, along with the number of heterozygous or homozygous individuals. In the present study, these variants were sys-

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†The authors wish it to be known that, in their opinion, the first two authors should be regarded as joint First Authors.

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## Research Article

## Pharmacogenomics



## Pharmacogenomic landscape of COVID-19 therapies from Indian population genomes

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**Aim:** Numerous drugs are being widely prescribed for COVID-19 treatment without any direct evidence for the drug safety/efficacy in patients across diverse ethnic populations. **Materials & methods:** We analyzed whole genomes of 1029 Indian individuals (IndiGen) to understand the extent of drug-gene (pharmacogenetic), drug-drug and drug-drug-gene interactions associated

RESEARCH

Open Access



# Genetic epidemiology of autoinflammatory disease variants in Indian population from 1029 whole genomes

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## Abstract

**Background:** Autoinflammatory disorders are the group of inherited inflammatory disorders caused due to the genetic defect in the genes that regulates innate immune systems. These have been clinically characterized based on the duration and occurrence of unprovoked fever, skin rash, and patient's ancestry. There are several autoinflammatory disorders that are found to be prevalent in a specific population and whose disease genetic epidemiology within the population has been well understood. However, India has a limited number of genetic studies reported for auto-inflammatory disorders till date. The whole genome sequencing and analysis of 1029 Indian individuals performed under the IndiGen project persuaded us to perform the genetic epidemiology of the autoinflammatory disorders in India.

**Results:** We have systematically annotated the genetic variants of 56 genes implicated in autoinflammatory disorder. These genetic variants were reclassified into five categories (i.e., pathogenic, likely pathogenic, benign, likely benign, and variant of uncertain significance (VUS)) according to the American College of Medical Genetics and Association of Molecular pathology (ACMG-AMP) guidelines. Our analysis revealed 20 pathogenic and likely pathogenic variants with significant differences in the allele frequency compared with the global population. We also found six causal founder variants in the IndiGen dataset belonging to different ancestry. We have performed haplotype prediction analysis for founder mutations haplotype that reveals the admixture of the South Asian population with other populations. The cumulative carrier frequency of the autoinflammatory disorder in India was found to be 3.5% which is much higher than reported.

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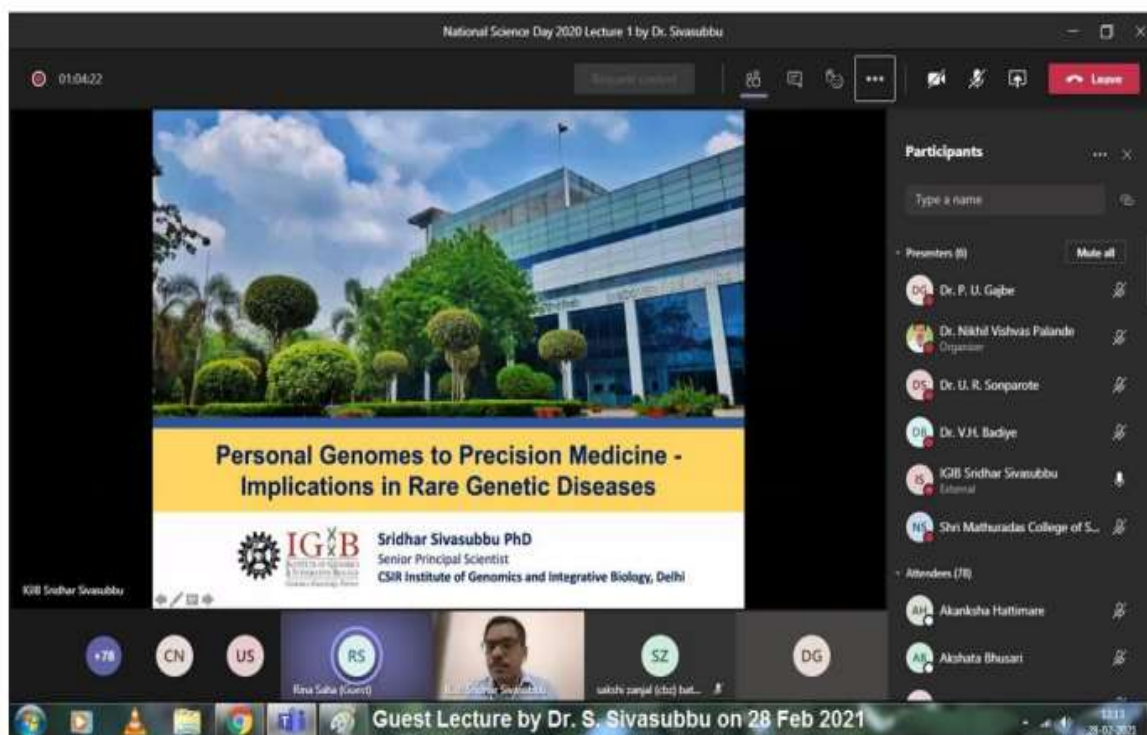
Full list of author information is available at the end of the article



### Activity/outcome 2 of MOU No. 1

2 online guest lecture was arranged under this MOU on 28<sup>th</sup> February 2021 and 02 March 2021 as part of the **National Science Day celebration**.

- **Guest lecture 1**



Presentation of National Science Day on 28<sup>th</sup> February 2021

**Report-** Guest Lecture on the topic “Personal Genomes to Precision Medicine-Implications in Rare Genetic Diseases”

**Mode-** Online

**Name of Guest-** Dr. Sridhar Sivasubbu

**Date-** 28<sup>th</sup> February 2021

**Venue-** Online

**Brief Report:** National Science Day was celebrated by organizing an online Guest Lecture on the topic “Personal Genomes to Precision Medicine-Implications in Rare Genetic Diseases” by Dr. Sridhar Sivasubbu, Senior Principal Scientist, CSIR Institute of Genomics and Integrative Biology, New Delhi.

- Guest Lecture 2



Presentation of online Guest Lecture by Dr. Vinod Scaria on 02 March 2021

**Report-** Guest Lecture on the topic Covid-19 Genomics: Mutations, Reinfections, and Escapes

**Mode-** Online

**Name of Guest -** Dr. Vinod Scaria

**Date-** 2<sup>nd</sup> March 2021

**Venue-** Online

**Brief Report:**

Guest Lecture by Dr. Vinod Scaria, Principal Scientist, CSIR Institute of Genomics and Integrative Biology (IGIB), New Delhi was organized. His presentation was on the topic “Covid-19 Genomics: Mutations, Reinfections, and Escapes”.

