DEPARTMENT OF BIOTECHNOLOGY

EXTENSION ACTIVITIES

2020-2021

Programme I

In session 2020-2021, the Sickle Cell Anaemia related extension programme has been carried out at Kawardha district of our state. We followed same protocol as our continuous programme.

Fig. showing Sickle Cell Anaemia Screening Programme organized at Chandaini village of Kawardha District of Chhattisgarh.

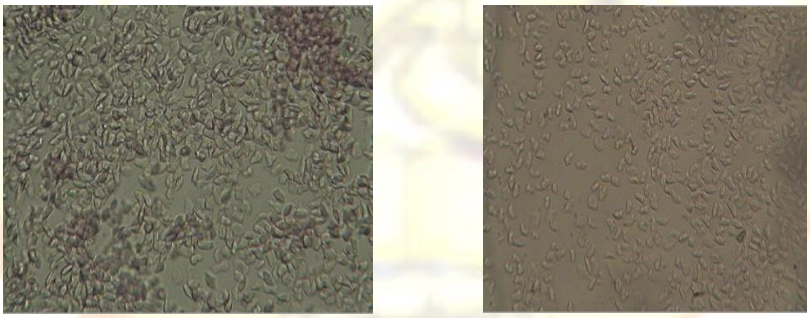

Fig. Showing sickle-shaped RBCs.

- **Programme II**

The Second continuous extension programme of the department i.e., Glucose-6-Phosphate Deficiency and related genomic analysis from the society has been carried out at Kawardha district in session 2020-2021. It was on the same line of previous year programme.

Fig. showing sample collection at Government H.S. School Bodla, Kawardha district of Chhattisgarh.

- **Programme III**

Chemokine receptor gene frequency analysis has been extended this year also but from the population of Kawardha district with same aim and objective.

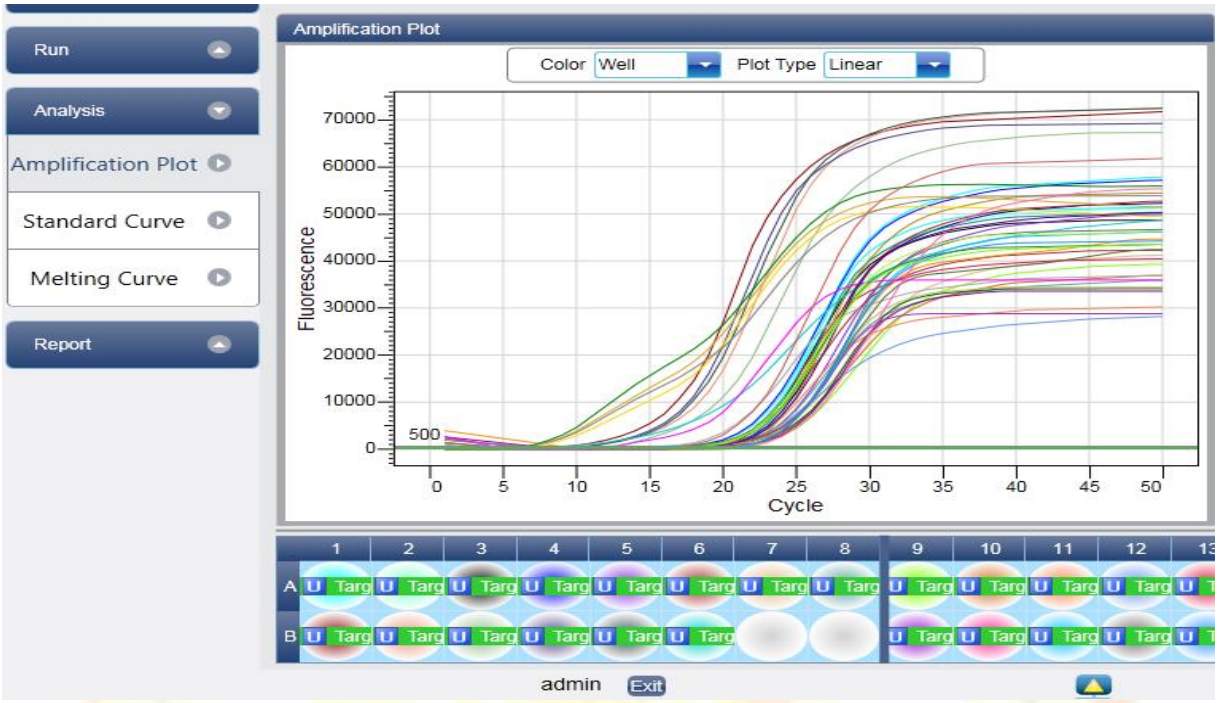

Fig. Showing expression of receptor genes for Chemokines.

- **Programme IV**

In our state one village Supebeda of Gariyaband District is worst sufferer of Renal dysfunction and the reason is unknown. Out of 1200 population, nearly 150 have died and our government is seriously concerned for this problem. We are analysing heavy metals from blood & urine of sufferer and analysing their genome to detect cause of the problem to serve society and to help medical department and government. AIIMS Raipur is our collaborator in this work.

Fig. showing sample collection at Village Supebeda of Gariyaband District of Chhattisgarh in collaboration with AIIMS Raipur.

3billion

Report date: 2021-12-06

RARE DISEASE GENETIC TEST

RESEARCH USE ONLY

Unique ID: MDSC-15

Sex: Female
Sample ID: EPJ21-AABC
Date of birth: 2000-05-11
Ethnicity: South Asian

ORDER INFORMATION

Physician Information

Name: .Anil Kumar
Medical speciality: -
Email address: aimum_aishley@yahoo.co.in
Phone: +91 98274-91253

Institution Information

Name: Government V.Y.T.PG Autonomous College
Address: G.E.Road Durg Chhattisgarh India

Order Information

Test: Whole exome sequencing
Product type: Proband only
Specimen type: EDTA blood
Order date: 2021-10-19
Sample collection date: 2021-10-17

TEST RESULT

Inconclusive — A pathogenic variant was identified.

A pathogenic variant was identified in HBB gene that may explain the following patient's phenotype: sickled erythrocytes.

Gene	Variant	Classification	Disease
HBB	11-5248232-T-A NM_000518.5:c.20A>T (NP_000509.1:p.Glu7Val) Heterozygous	Pathogenic	Sickle cell anemia

Interpretation

The patient's phenotype is considered compatible with Sickle cell anemia, which is an autosomal recessive disorder. However, because only a single heterozygous variant was detected, a molecular diagnosis cannot be established at this time. Other genetic testing may be able to identify the second variant such as deletion, duplication and deep intronic variant that is not detectable by this test.

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RESULT INTERPRETATION

HBB NM_000518.5:c.20A>T (NP_000509.1:p.Glu7Val)

Population frequency	It is observed in the gnomAD v2.1.1 (https://gnomad.broadinstitute.org/) dataset at total allele frequency of 0.00348.
Variant type and location	None
Case-level data	The same variant was observed in multiple affected individuals with a consistent phenotype from unrelated families (PMID: 25023084, 25203083, 25023085). This variant was previously reported in trans with another pathogenic variant in this gene (PMID: 23591685, 29542687).
Functional and computational data	Functional assays showed that the variant had strong level of impact on gene/protein function (PMID: 1802884, 2296310, 28356267, 12124399). In silico prediction tools and conservation analysis predicted that this variant was probably damaging to the protein structure/function (3CNET: 0.83>=0.75).
Association with known pathogenic variant	Amino acid change identical to known pathogenic variant has been previously reported with established evidence (ClinVar ID: VCV000015333, PMID:3267215). Different pathogenic amino acid change has been reported with sufficient evidence at the same codon (ClinVar ID: VCV000015126,VCV000036301, PMID:19460936,6129204,8294201).
Relevance to disease	Sickle cell anemia
Validation	Not performed as the variant was high-quality
Conclusion	Pathogenic

Sickle cell anemia (OMIM: 603903)

Sickle cell anemia, associated with HBB gene, is an autosomal recessive disorder. Patients affected by the Sickle cell anemia present with pure red cell aplasia, persistence of hemoglobin f, increased red cell sickling tendency, bone marrow hypocellularity, increased mean corpuscular volume, chronic myelogenous leukemia, chronic hemolytic anemia, leukocytosis, microcytic anemia, hypochromic anemia, reticulocytosis, anemia, thrombocytosis, iron deficiency anemia, hemolytic anemia, priapism, sepsis, chest pain, cerebral palsy, pulmonary fat embolism, abnormality of pulmonary circulation, wheezing, night sweats, increased lactate dehydrogenase level, cough, pain, hypoxemia, fatigue, pigment gallstones, avascular necrosis, unconjugated hyperbilirubinemia, abnormal left ventricular function, poor appetite, elevated serum creatinine, respiratory failure, tachypnea, osteomyelitis, recurrent infections, recurrent bacterial infections, abnormality of the vasculature, hepatomegaly, pulmonary arterial hypertension, pneumonia, rigidity, abdominal pain, asplenia, splenomegaly, abnormality of the spleen, cardiomegaly, hepatic failure, cholestasis, stroke, intellectual disability, cholelithiasis, jaundice, osteoporosis, hypertension, hematuria, abnormality of the nervous system, retinopathy and renal insufficiency.

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Sex: Female | Date of birth: 2000-05-11

Sample ID: EPJ21-AABC | Ethnicity: South Asian

SECONDARY FINDING

No pathogenic or likely pathogenic variant detected in 73 medically actionable genes for secondary reporting recommended by American College of Medical Genetics (ACMG) Guideline (Genet Med. 2021 May 20).

TARGET REGION COVERAGE

	Target bp covered (%)				
Mean depth (X)	≥ 1X	≥ 5X	≥ 10X	≥ 20X	≥ 50X
138.10	99.2	99.0	98.9	98.8	96.6

METHODS

Genomic DNA was extracted from the blood, saliva, or buccal swab sample of a patient. All exon regions of all human genes (~22,000) were captured by xGen Exome Research Panel v2 (Integrated DNA Technologies, Coralville, Iowa, USA). The captured regions of the genome were sequenced with Novaseq 6000 (Illumina, San Diego, CA, USA). The raw genome sequencing data analysis, including alignment to the GRCh37/hg19 human reference genome, variant calling and annotation, was conducted with open-source bioinformatics tools and in-house software. The automatic variant interpretation software, EVIDENCE, was developed in-house to prioritize variants based on ACMG guideline (Genet Med. 2015;17:405-424) and the phenotype of each patient. This system has three major steps; variant filtration, classification and similarity scoring for patient's phenotype (Clin Genet. 2020;98:562-570). First, gnomAD (http://gnomad.broadinstitute.org/) as a population genome database and 3 billion genome database were used for estimating allele frequency. Common variants with minor allele frequency of >5% were filtered out in accordance with BA1 of the ACMG guideline (Genet Med. 2015;17:405-424). Second, we extracted evidence data on the pathogenicity of variants from a number of scientific literatures and disease databases including ClinVar (https://www.ncbi.nlm.nih.gov/clinvar/) and UniProt (https://www.uniprot.org/). Pathogenicity of each variant on its associated diseases were evaluated according to the recommendations of ACMG guideline (Genet Med. 2015;17:405-424). Third, the patient's clinical phenotypes were transformed to corresponding standardized human phenotype ontology terms (https://hpo.jax.org/) and accessed to measure the similarity (Am J Hum Genet 2016;98:490-9 and Am J Hum Genet 2009;85:457-64) with each of ~7,000 rare genetic diseases (https://omim.org/ and https://www.orpha.net/consor/cgi-bin/index.php). The similarity score between each patient's phenotype and symptoms associated with that disease, caused by prioritized variants according to ACMG guideline, ranged from 0 to 10. Finally, medical geneticists and medical doctors manually evaluate the candidate variants and associated diseases. Single nucleotide variants that do not meet our stringent WES quality metrics and all indels are confirmed using bidirectional Sanger sequencing.

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LIMITATIONS

1. Whole exome sequencing targets about 97% of the exon region in the human genome.
2. There are regions and genetic variants that cannot be technically covered by whole exome sequencing method.
   - Structural chromosomal aberrations including large copy number variation, translocation and inversion
   - Trinucleotide repeat expansion
   - Mitochondrial genome
   - Epigenetic factors
   - Low level mosaicism
   - Uniparental disomy
   - Variants in genes with corresponding pseudogenes or other highly homologous sequences, and non-coding regions including untranslated regions, introns, and intergenic regions
3. Results and interpretations were considered in context with clinical findings, phenotypes, family history of the patient. Genetic variations were reported only if they were relevant to the patient's clinical phenotypes. False interpretations may occur due to incorrect or incomplete clinical information reported for the patient. Additional genetic or non-genetic tests should be considered if results do not match the patient's clinical information.
4. Despite the daily update of our database on genes and diseases, the referenced information may not be up-to-date due to the constant addition of the new data. As the absence of reported pathogenic variants cannot be concluded that the patient's symptoms are not due to the genetic cause, we perform daily re-analysis until a diagnosis is made. Once a proper diagnosis is made, we will report it to the physician.
5. Sanger sequencing of biological parents is required for the segregation analysis on the identified pathogenic variants.
6. In the case of re-analysis or secondary finding, 3billion does not provide Sanger sequencing to confirm the identified variant.

This report has been reviewed and confirmed by our geneticists and physicians;

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Chief Medical Officer, M.D, Ph.D.

- **Programme V**

Our department was seriously concern for Air Quality Index influenced spread of COVID-19 epidemic, for that we have analysed and correlated the cases of COVID-19 with AQI of Chhattisgarh, Bihar and Madhya Pradesh and found NO_2 as most accelerating factor for hospitalization & need of ventilators of patients. We have also studied stress level among students during long lockdown period of epidemics.

Long-Run Dynamics of the Novel Corona Virus Infections COVID-19 concerning Air Quality Index, $PM_{-2.5}$, NO_2, PM_{-10}, and O_3 in the Chhattisgarh State of India

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Abstract

In this research, the long-run disease dynamics of the COVID-19 were studied concerning the Air Quality Index (AQI), $PM_{-2.5}$, NO_2, PM_{-10}, and O_3, respectively, by using eigen space decomposition. Change in COVID-19 related to AQI showed that initially when the AQI changed from 103 to 84.83 the disease dynamics also changed, and the first cases of COVID-19 were reported. In the next two fortnights from March 15, 2020, and April 01, 2020, the dynamics were the same, latter the AQI changed from 84.83 to 63.83, but this change does not affect the disease dynamics in long run from April 15, 2020, to Jul 15, 2020. In Phase-1 the time duration was from March 15, 2020, to May 01, 2020, and for Phase-2 the time duration was from Jun 01, 2020, to Jul 15, 2020. In phase 1 the solution obtained shows a cyclic trend with initially decreasing, then increasing, and again a decreasing trend for changes concerning $PM_{-2.5}$. The disease dynamics concerning $PM_{-2.5}$, NO_2, PM_{-10}, and O_3, respectively, based on initial transition showed the same trend for $PM_{-2.5}$, NO_2, and PM_{-10}. Moreover, for O_3 the disease dynamics were found different than the other three parameters. The findings of the present study prove that the Eigen Space Decomposition method is one of the significant tools for planning control measures for disease with compatibility to air quality alterations.

Keyword: Novel Corona Virus, AQI, $PM_{-2.5}$, NO_2, PM_{-10}, O_3, Eigen Space Decomposition, COVID-19

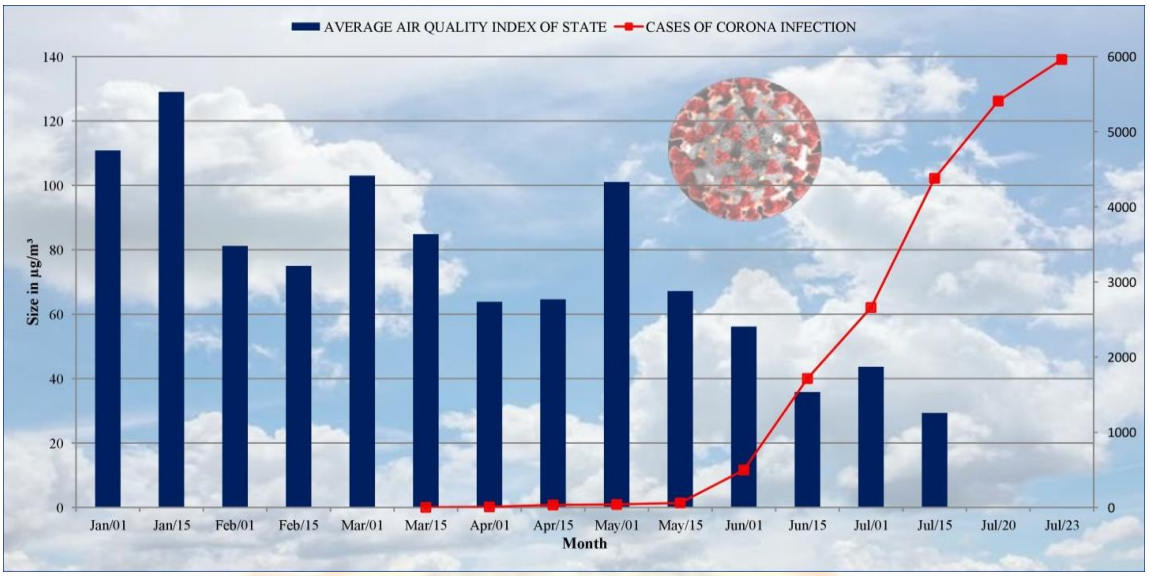


Fig. showing a negative correlation between air quality index and surge of COVID-19 has been reported from Chhattisgarh.

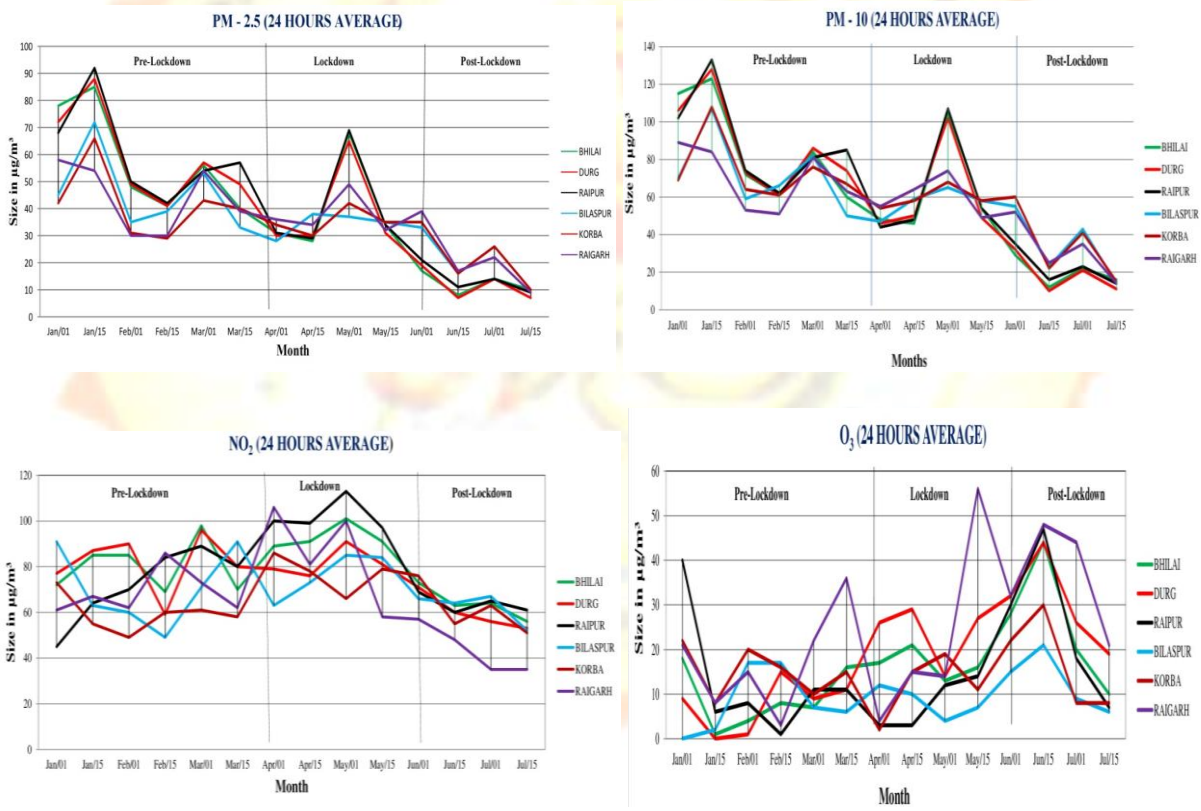


Fig. showing unprecedented surge in NO2 has been found during lockdown period in Chhattisgarh which accelerated lungs dysfunctions during COVID-19 epidemics

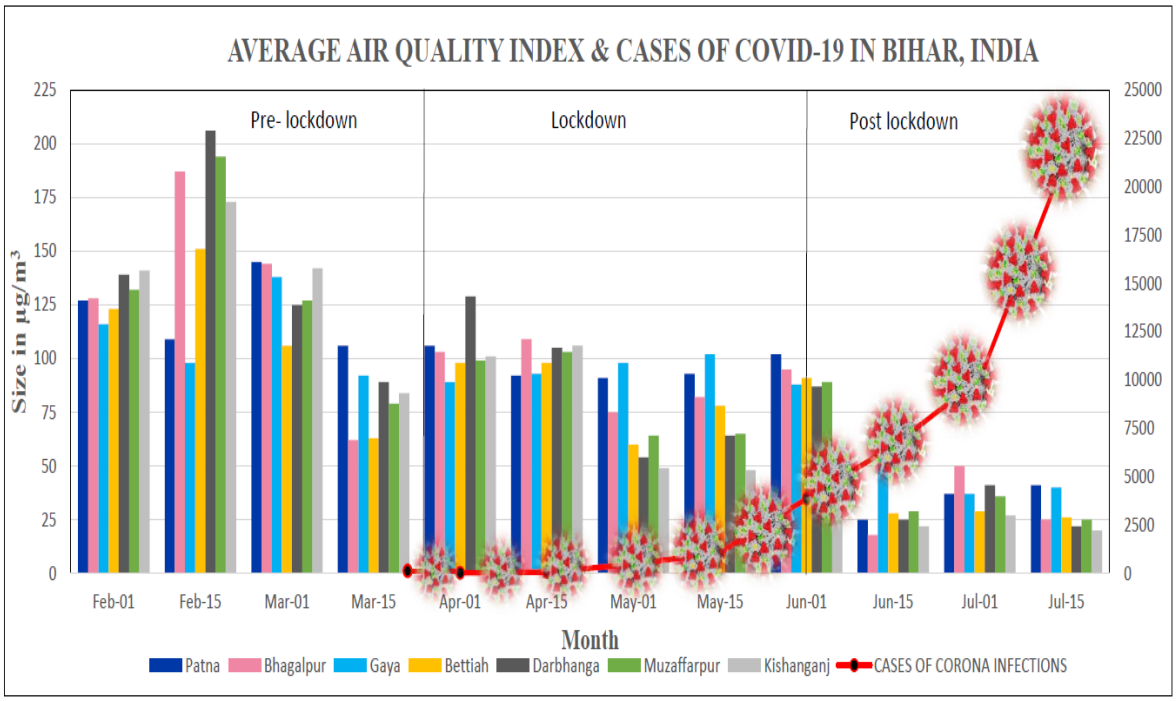


Fig. showing AQI Analysis of Bihar State during Lockdown period

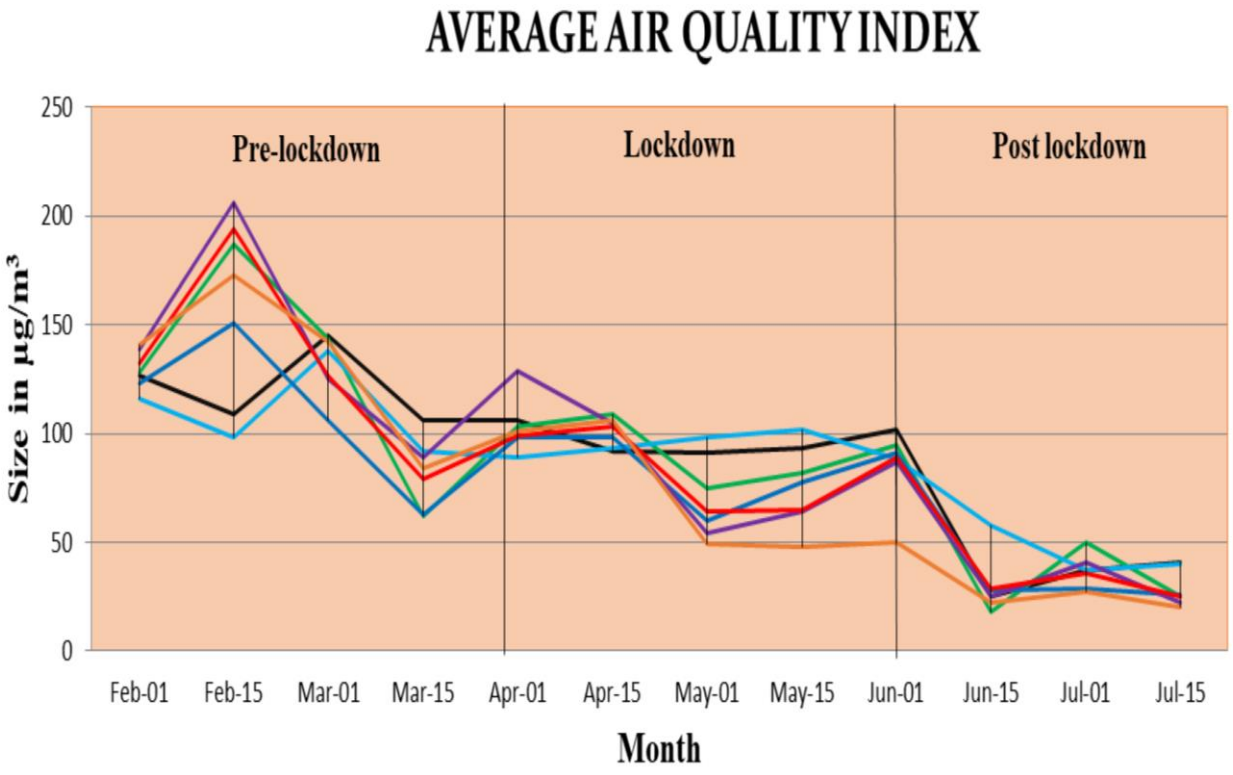
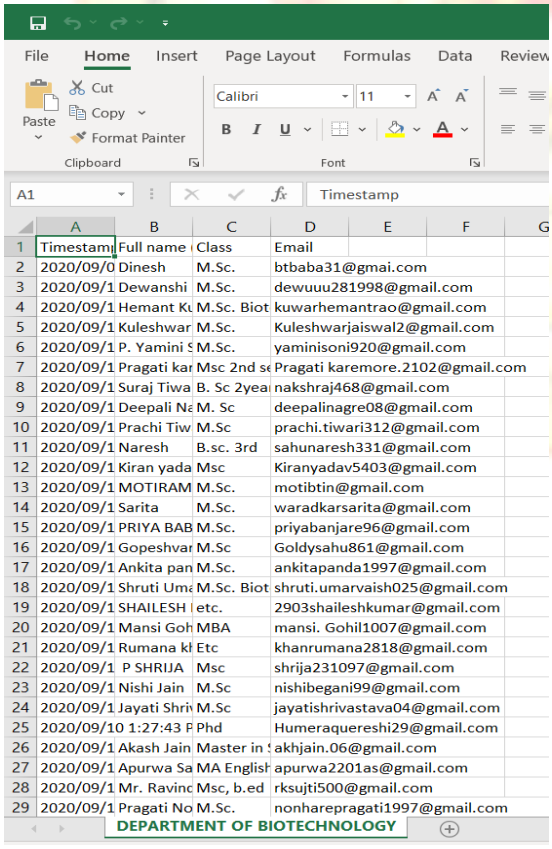


Fig. AQI Analysis of Madhya Pradesh State during Lockdown period.



Timestamp	Full name	Class	Email
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2020/09/1	Dewanshi	M.Sc.	dewuuu281998@gmail.com
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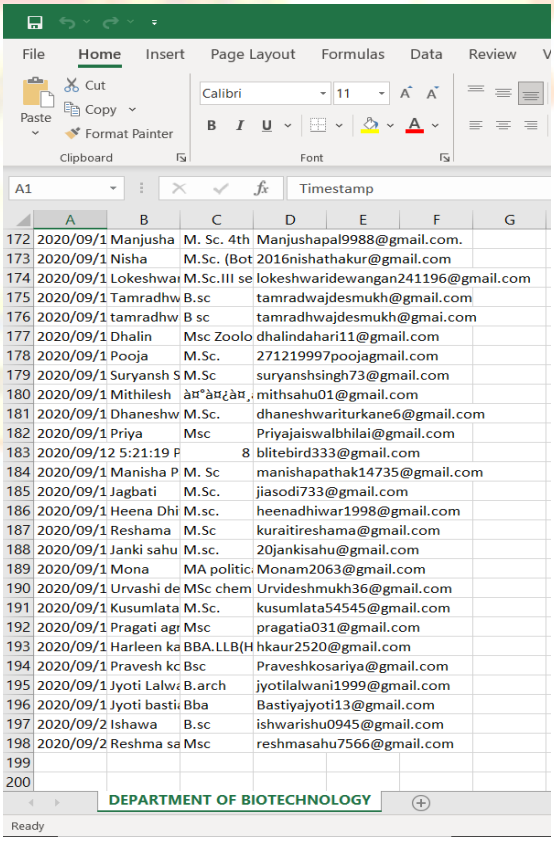

Timestamp	Full name	Class	Email
2020/09/1	Manjusha	M. Sc. 4th	Manjushapal9988@gmail.com.
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2020/09/2	Reshma sa	Msc	reshmasahu7566@gmail.com

Fig. showing registration of students for Impact analysis of lockdown on mental stress level.

- **Programme VI**

The environment analysis of Shivnath river from Durg to Rajnandgan has been continued this year also for its physio-chemical analysis and biological analysis with special reference to Molluscans and aquatic insect diversity. The purpose was to know the impact of environmental alteration and its adverse impact on human population of catchment area.

Fig. Showing pollution status of Shivnath River

Fig. Showing Insect collection from Shivnath River