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CLOUD-BASED DIGITAL PLATFORM FOR GENETIC LAB WITH PREDICTIVE ANALYTICS & REPORTING USING AI

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Abstract— *In this day and age, computers have become a necessity in nearly every field and industry. The 21st century witnessed a digital revolution and realized the enormous benefits of utilizing digital technologies over traditional methodologies with respect to convenience, speed and accuracy. In the healthcare industry, digitization is used to leverage patient empowerment and adherence to various treatments. Cloud Based Digital Platform for Genetic Lab is a platform which is designed to digitize different processes and treatments involved in the workflow of a Genetic Laboratory. It is basically a Greenfield Digital Transformation of a genetic testing lab at every single operating point and an end to end new-age digital platform. It is a bespoke digital framework that provides high availability, security, interoperability, versatility and digital agility. There are multiple products in this ecosystem which basically talks to a digital platform. The idea of this project is to build a robust platform to manage various entities that interact with the lab such as the institute, doctor, patient, counselor . The platform applies machine learning models for predicting anomalies and to aid in the detection of any disease or deficiency in prenatal genetics and aims to use supervised learning models to build specific AI in the field of genetic reporting & anomaly detection. The platform allows the patients to get digital consultations, treatments, reports and accurate insights, thereby increasing their convenience and satisfaction.*

Keywords — digital transformation, healthcare, genetics, genetic correlation, predictive analytics

I. INTRODUCTION

The field of Genetics is poised to become the cornerstone of medicine as we know it. In the decades to come, we will witness a startling transformation in the way we manage diseases and their therapies. Much of this transformation will account due to ground breaking advances in clinical and human genetics happening today. This has opened the doors to new business opportunities that lie completely untapped and unseen. One such opportunity is genetic diagnosis & testing for mutations & genetic anomalies. The genetic testing lab under consideration conducts the following modalities:

1. Reproductive Genetics
2. Cancer Genetics
3. Genetic Consultation & counseling
4. Metabolic Genetics
5. Fetal Autopsy

A. Executive Summary

The field of Reproductive Genetics deals with conducting various specialized genetic screenings to assess the risk of a fetus, newborn baby or an individual (mostly couples) suffering from genetic

abnormalities, disorders & diseases. Genetic screening, testing & diagnosis differs from ordinary pathology in not just the process, expertise & technology required, but the resultant reports are not simple computer outputs, hence require expert intervention & interpretation.

B. Theoretical Framework

The project is broadly broken down into following applications: Lab Digital Platform, Customer facing platform, consumer facing platform and Analytics platform. The digital platform is the cornerstone of this digital transformation project. It houses the central data warehouse, data lakes and machine learning models. The customer facing applications help Hospitals, Doctors, Doctor Assistants and other medical professionals to execute their professional duties and tasks.

II. Scope

A. Justification

Through the Digital India campaign, many sectors have converted into true digital states. Healthcare is a sector that a large percentage of the public do not approve to be digitized. But in this evolving world it is a necessity. Digitization not only unburdens various tasks by automation, but also increases efficiency, enforces safety, increases storage, integrates business and resources and eliminates error rate.

B. Product Scope Description

The platform is supported on all major operating systems such as iOS, Android, Windows, etc with different user specifications and automates the process from filling the TRF to report generation and analytics using a centralised cloud database system for securely storing and retrieving sensitive information.

C. Acceptance Criteria

The platform should be able to accept details and strictly validate them using complex algorithms to ensure that the entered information is authentic and sensible. It should be secure and robust for protecting sensitive information of patients. It should allow basic functions such as booking an appointment for genetic testing or consultation. It should let doctors prescribe tests, view appointment details and manage and publish test reports after proper assessment. The different entities such as Institutes, Doctors, Doctor's Assistants, Patients, Counsellors and Franchise must be managed using the platform.

D. Deliverables

The platform is able to deliver the entire genetic testing process on any smartphone or web application where they could process the patient's information securely. Each patient record undergoes complete registration as well as being confirmed from the patient side. The information which is collected and stored on a cloud based storage makes the system more efficient and which also benefits for the analysis purpose.

E. Assumptions

1. Institute is expected to be government recognised and should have a valid certificate for conducting

genetic tests.

2. All entities i.e the end user of the application have the prior knowledge of the entire process.
3. The end user should have an android or iOS smartphone or a browser to access the Web App.
4. The end user is aware of the existing workflow of the organization.
5. The end user has the appropriate and legal credentials to access the web app.

III. Literature Review

Digitization of healthcare data in a manner that is easy for computers to utilize is important to support the delivery of care through data visualization, collaboration, and clinical decision support. When data is digitized, we can create new and useful ways to display and visualize this data, which has the potential to provide better insights into a patient's status and, optimistically, better decisions. Another important application for digitized healthcare data is clinical decision support (CDS).

CDS systems combine the data with clinical knowledge to provide patient-specific suggestions at the appropriate time by

- Identifying at-risk patients;
- Tracking clinical outcomes;
- Performance measurement and management;
- Clinical decision making at the point of care;
- Length of stay prediction;
- Hospital readmission prediction

Hermes, S., Riasanow, T., Clemons, E.K.

On average around 75,000 preventable deaths occur each year in the United States and that health information technology (HIT) is a promising solution to this problem. Also, HIT has been recognized as a driver of enhanced clinical outcomes and as a cost-saving lever, yet adoption of health IT is slow. Which indicates HIT is resourceful in about saving 1% souls i.e fetus, due to abnormality or genetic cause.

V. Tresp, J. Marc Overhage, M. Bundschus, S. Rabizadeh, P. A. Fasching and S. Yu

For decades, much of what was documented about a patient was in paper format and collected in a folder that was physically moved across the clinical departments and was eventually filed. Today, patient data is increasingly recorded and stored in an electronic form, the EHR. The EHR greatly improves the quality of the data documented and supports improvements in patient care by enabling analysis and decision support. EHR consists of the same paper documents except that they are scanned and stored digitally. A significant amount of data must be entered by providers. Because of the time and effort required for providers to capture structured data, they often question if there is sufficient value to warrant the negative impact on productivity. Deidentification is the process used to prevent a person's identity from being connected with information. Common uses of de identification include human subject research, which requires privacy protection for research participants.

The healthcare sector and generally many other areas such as transportation, finance industry, etc. have gone through a rapid growth recently due to the exponential growth in ICT. The increasing role and benefits of ICT in healthcare are becoming visible in the enhancement and emergence of technologies such as health informatics, epidemiology, bioengineering and Healthcare Information Systems (HIS).

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Healthcare providers can port powerful analytics and decision support tools to mobile computing devices (smartphones, tablets, laptops, etc.) aiding clinicians at the point of care helping them with synthesis of data from multiple sources, optimization of clinical workflows, and context-aware decision making.

Table I
Summary Table Of Previous Researches

Sr.	Year	Author	Research summary	Remarks
1	2018	Belliger, Andréa & Krieger, David	Healthcare is no longer primarily something that takes place in the intimacy and confines of the doctor-patient relationship. Instead, health care is distributed throughout a complex network of both human and nonhuman actors such as databases, hospital information systems, digital health records, electronic health cards, online patient communities, health related apps, smart homes with ambient assisted living technologies, etc.	Services are more inclined towards technology, as in present scenario in Digital India
2	2020	L. Ivančić, L. M. Glavan and V. B. Vukšić	Digital transformation has reached out to every aspect of human lives, including personal and institutional provided healthcare. A systematic literature review and search in the Clarivate Analytics' Web of Science database was carried out, along with the text mining in order to analyze the papers.	
3	2020	Sascha Kraus, Francesco Schiavone, Anna Pluzhnikov, Anna Chiara Invernizzi	Prior research falls into five clusters: operational efficiency by healthcare providers; patient-centered approaches; organizational factors and managerial implications; workforce practices; and socio-economic aspects. These clusters are linked together into a model showing how these various forms of technology implementation lead to operational efficiencies for services providers.	
4	2020	Hermes, S., Riasanow, T., Clemons	States the digital platform exercise on well situated company and developed e3 value method to engage digital reforms in healthcare system	

5	2016	V. Tresp, J. Marc Overhage, M. Bundschus, S. Rabizadeh, P.A. Fasching and S. Yu	Indicates the pattern of digitalization in healthcare and large-scale data analytics. Tackle the movement toward continuous healthcare where wearable and stationary devices control health. Patients are gradually taking responsibility for their own health records.	The openness and transparency increases trust, thus portraying reliability.
6	2018	L.A. Tawalbeh and S. Habeeb	A structured healthcare approach to evolving principles of urban developments such as smart cities will manage health for the entire population at the region, country or global level. In future healthcare systems, mobile computing will play an integral role as mobile devices provide an interface between patients and healthcare practitioners and organisations.	
7	2019	S. Chakraborty, S. Aich and H. Kim	The primary concern in the Smart Healthcare scenario is the privacy and protection of patient's data. This privacy issue and also mitigation of accurate data has been very much handled in the work by controlling, tracking and sensing a paradigm as a transaction and access management framework in compliance with the IOT and the Blockchain. An effective forum for correct and trustworthy information to be used for deliberate medical treatment and the benefits of patients.	
8	2020	A.A. Ansari, S. Chakraborty, S. Aich, B. S. Kim and H. Kim	Optimize the healthcare network by allocating and distributing healthcare services as required to maximize service quality.The average fraction of population distribution of healthcare services is rendered from the viewpoint of age group and economy. The proposed effort is to assist the Ministry of Health in amending, expanding and strengthening the personnel and services of existing facilities.	
9	2020	M. G. do Nascimento	Ongoing pandemic COVID19 has extensively increased the utilisation of existing technologies like big data, recommendation systems, cloud computing and basic IOT.	Digitization is important in contactless service delivery.

10	2019	M.Chakraborty and K. M. A. Khan	Case study on a Hospital which adopted HRIS (Human Resource Information System) which proved to sustain work balance in the institute in a profitable manner.	HRIS has proved to be a powerful tool in the healthcare sector.
11	2018	Fuchs, Christoph & Hess, Thomas.	Emphasis on large scale agile digital transformation and records of its challenges and action. It includes interviews and case study of two major high revenue industries and depicts the agile journey for the same.	

There exist many other forms of processing data for health care services like MCC. Mobile Cloud Computing (MCC) integrates mobile devices to utilize the cloud unlimited service and to enable information access for mobile devices users. The Cloud Computing depends on network-connected resources pooled to maximize their utilization resulting in reduced management and capital costs. Many sectors can benefit from MCC including the cloud-healthcare system. As an example, there is a MCC healthcare system that was built to observe and analyze real time biomedical signals (such as ECG) for users in multiple locations. A personalized healthcare application is installed on the mobile device, and health data is being synchronized into the healthcare cloud computing service to be stored and analyzed.

The traditional methods or systems that are available for genetic lab include local data entry of Transfer Requisition Form (TRF). A hard copy is filled by the patient or an assistant with their personal information like name and contact details which in turn is converted into soft copy via means of direct typing into a software integrated database. Further processes are carried manually that includes testing, concluding and report processing.

Limiting factors include the following:

1. These systems need human interference for information exchange.
2. In light of filling TRFs, method is time consuming and labour cost increases overall capital.
3. No system exists for institutes like hospitals to access the lab system directly, without human interference.
4. There is no such system where patients could have direct interaction with the laboratory on a digital platform.
5. A local system failure results in the flow of the system.

IV. Proposed System

The proposed system aims at digitizing and transforming the operations for a genetic lab wherein the entire laboratory will work on a digital platform. As this is not a normal laboratory in which the samples are tested and report is generated rather this lab works with those samples that are of very high importance and requires utmost care. However, the sample information must be kept confidential as these tests are permission of government authorities.

The main aim is to make use of the existing traditional approaches and provide a more simpler, centralized and automated approach. The paperwork done in the laboratory could be digitalized keeping in mind the importance of sample information.

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The entire system can be understood better by the following modules:

A. Databases

The database is the most fundamental part of the application as the entire record maintenance will be on the cloud, more precisely, a centralised cloud. The database architecture for each operating app is separate with some exceptions where an employee works in more than one organisation. The database for users will be centralized for reasons such as reducing latency while logging in this also helps the switching of operating companies and/or domains easily.

All the apps will be interacting with the centralized database for getting the things from one end. All the confirmation within each entity will be updated in the database and entities would be notified, once the data is updated. Obviously, the generated database would be large and the data will be continuously getting updated in the database, along with which the analysis of the data would be also done simultaneously and would be getting displayed in the dashboard of some apps.

Considering both the points the database technology that will be used is MongoDB. As it stores data in the form of documents, storing and analyzing both becomes relatively easy and simple. Along with giving us the benefits we need it easy to scale and relatively easy to work with once the engine is properly configured.

B. Applications

There is a separate application for every entity. In total, there are eight applications that are supported on all commonly used operating systems.

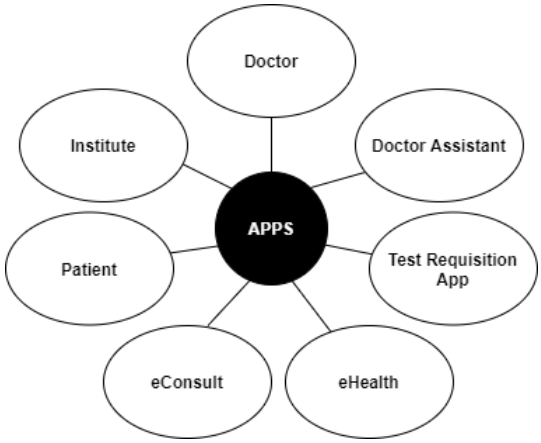

Fig. 1: Apps

1) *Institute App:* Institute is the business entity under which doctors, hospitals, nursing homes and polyclinic hospitals are mapped. The Institute app is used to manage doctors, hospitals, nursing homes and polyclinic hospitals. Operations such as sample collection, payments, reporting, etc are included on the app.

2) Doctor App: Doctor app will be used by doctors for all their medical / scientific needs & services beyond those. Each doctor is mapped to an institute. Essentially, an institute is the business entity under

which a doctor is mapped as a practitioner. Hence one doctor may belong to multiple institutes & one institute may have multiple doctors under it. It is not possible for a doctor to exist or order tests without being mapped to an institute. There can be their own institute that will be created as a default requirement.

3) *Doctor Assistant App:* Doctor assistant app will be used by doctor's assistants who will perform the role of supporting all of the doctor's needs like test ordering, follow-up on report, patient engagement wherever necessary. The difference between a doctor assistant & doctor is that the assistants are qualified medical representatives (BHMS, BAMS, BUMS, tc) while doctors are highly qualified specialized doctors (MD, MS, etc). Doctor assistants are first mapped to doctors, then to institutes. In other words, one assistant can be mapped to multiple doctors within an institute. At the same time, one assistant may be mapped to one doctor who visits different institutes.

4*) Test Requisition App :* Test Requisition App app is the core module of the entire platform that will be used for ordering tests, completing the mandatory process of TRF (Transfer Requisition Form) fulfilment. Each Test Requisition App user login is mapped to an institute. Essentially, Test Requisition App is owned by an institute & will be used by the institute authorized staff for test ordering. The process starts when a doctor prescribes a test to a patient. After that, the authorised institute staff or the doctor (or the patient if no medical assistance is required) starts filling up the Test Requisition App that consists of the following 7 steps:

I. Doctor's details: This includes the type of doctor and depending on that, another field(s) will be displayed. For eg, if a Gynaecologist is selected, then the name of the Sonographer has to be selected as well. There is also an optional field for specifying the referring doctor.

II. Details of the Test(s): Here, the different types of tests that are needed to be performed are entered here. There is also a validation that makes sure that the tests don't contradict each other. For eg, if a test that is performed during the first trimester is added to the list, then a different test that is performed in the second or third trimester could not be added due to obvious reasons.

III. Collection Location: This could be either done at the Institute or Non-institute location.

IV. Patient's Details (KYC): The name of the patient is entered/selected depending on whether the patient is existing or a new one and the person (staff or patient) who is going to fill the form is specified. If staff is selected, then the fields that accept personal, contact and physical information appear. Whereas if the patient is selected, then a link is sent to the patient's phone number for completing the KYC.

V. Clinical Information: This is the most complex part of the form. This part partially depends on the test(s) that have been selected in step 2. It covers many specific details that are necessary for performing the test and also the scans that are required. In the end, the sample info is entered that includes the container ID and its type.

V. *Payment:* The payment could be made towards the institute or the lab. If the lab is selected, then payment type (online or offline) is asked, whereas if the institute is selected, then the institute is

responsible for it. After that, the entity who will be confirming the details about the test is to be specified. It could be either the staff or the patient.

VII. Confirmation: If the staff is confirming the details, then the page containing the information is displayed and if the patient is doing it, then a link would be sent to the patient so that the patient can verify their details and accept or reject the Test Requisition App. The staff can submit the Test Requisition App and it gets displayed on the dashboard.

5) *eHealth App:* eHealth app will be used by counselor and counselee for digital consultation. Through this app, counselors can provide digital consultation for emotional / physiological and medico-legal problems to patients.

6) *eConsult App:* eConsult app will be used by counselor and counselee for digital consultation. Through the eConsult app, counselors can give general medical, genetic & clinical consultation. The difference between eHealth and the eConsult is that the counselors can refer for a test through the eConsult app.

7) *Patient* App: Patient app is the application that will be used by patients (customers) for booking appointments and digital counselling.

VI. Design

A. Selection of the Technology Stack

As this system is required to be supported on Android, iOS, Linux, Mac, Windows & the web, the best framework to use for the frontend would be Flutter, as it is used to develop cross-platform applications from a single codebase. Having a single codebase allows you to use only one programming language for developing applications that will be supported across all platforms.

For the backend, NodeJS was selected due to its event-based model, scalability, rich ecosystem and robust technology stack.

For the database, MongoDB was chosen because it is able to handle & process large amounts of data very quickly. It is a NoSQL database which means that it doesn't have a fixed schema that might result in scalability & performance issues.

B. The Process

The platform is designed in a way that makes the entire process convenient and streamlined by eliminating the repetitive steps at every single operating-point. The process starts when a patient books an appointment and approaches the doctor for medical consultation. Both the patient and doctor are required to be registered in the system. The patient may be registered upon first visit to the doctor or while filling the Test Requisition App. The doctor will diagnose the patient and, based on the diagnosis, prescribe some test(s) for further investigation.

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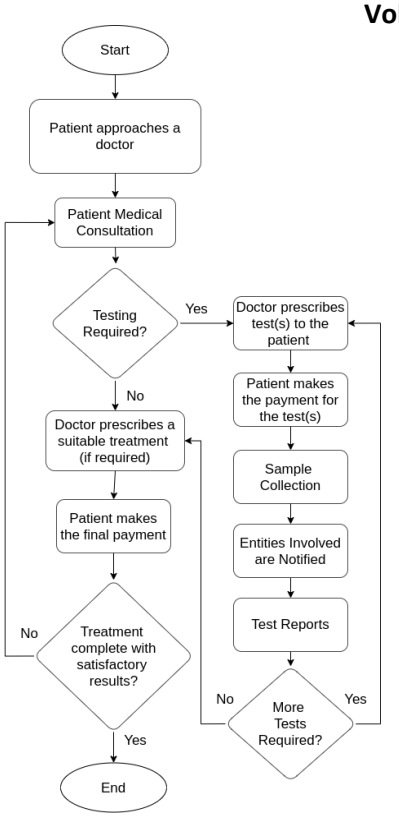

Fig. 2: Process Overview

These tests are then redirected to the institute authorized staff which will fill up the Test Requisition App form according to the doctor's prescription and after the payment has been made by the patient, the required samples will be collected from the patient either at the institute or some other suitable location. The sample collection process takes care of all the different scenarios that might be problematic. Sometimes, there could be an immediate need for a sample in case of an emergency. The doctor's assistant might not be available and so the doctor needs to take care of those tasks.

After the sample has been collected successfully, it is registered on the platform and the information is sent to the laboratory where the sample will be tested and the reports will be generated. These reports are then made available to the doctor who will then analyse the reports and give their diagnosis and opinion on it. If further clarifications are required, the doctor would prescribe more tests to the patient.

The system is built to derive meaningful insights from the tests and correlate the information with other similar instances to help in making the right decision. These decisions are extremely critical especially when it comes to prenatal genetics. Based on these decisions, the doctor is required to prescribe proper treatments to the patient to achieve the desirable outcome.

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VII. Abbreviations

- HIT - Health Information Technology
- EHR - Electronic Health Record
- ICT - Information & Communications Technology
- MCC - Mobile Cloud Computing
- ECG - Electrocardiography
- HIS - Healthcare Information Systems
- Test Requisition App - Digital Transfer Requisition Form

VIII. Conclusion

Health is something that cannot be bought or sold to anyone. It is priceless and it should be treated with utmost care. This project aims at improving and saving lives by providing a platform that could be trusted by doctors as well as patients for deriving meaningful insights related to their health and genetics. These insights will be of enormous value as they will be utilized to make extremely critical clinical decisions that might affect an individual's life for the better. This is just a small stepping stone towards a healthy and digital future.

IX. Future Scope

The scope of medical science has no end until life exists on the planet. The medical field can be integrated with many domains, thus the project has a huge scope in the future. The advancement in the genetic field has become a growing industry in today's era.

A. Integrating Deep Learning for Making Complex Predictions

As critical clinical decisions are involved, one can make use of deep learning with further research on the subject for performing more accurate & complex predictions related to genetics for the laboratory.

B. Implementing the System Across Multiple Laboratories

The methodology implemented for this project can be generalized to many other genetic and non-genetic labs for advancing their procedural tests being conducted in their laboratory. This idea can be extended for conducting general tests like blood sampling, sugar monitoring, etc. and storing the results for making well-informed decisions in the future.

C. Streamlining the Process Using IoT

One can integrate the platform with IoT devices which may be as simple as doing biometric tests or it could be combined with the lab instruments for reducing human interventions thereby eliminating human errors.

D. Secure Digital Consultation through Video Conferencing

The digital consultation process can be made more personalized by allowing the option to conduct video conferencing between the counselor & the patient as text-based communication can reveal only limited information about the patient.

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