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EXECUTIVE SUMMARY

Using a combination of literature surveys and stakeholder interviews, the authors evaluated India's rare disease landscape and the barriers to enabling patient-owned health data registries for national and international data sharing.

While there is poor understanding among the general public about rare diseases, newer genetic technologies for diagnosis as well as the benefits of data sharing, different stakeholders have expressed interest in participating in a patient-owned rare disease registry and the main concerns raised were regarding data privacy and social stigma around rare diseases.

India currently has very few standardized registries, which are largely not patient-owned. India also does not have a Rare Disease Policy and there are no specific guidelines or regulations regarding collection and storage of data pertaining to patients with rare diseases. However, the country is moving towards digitizing healthcare with the newly launched National Digital Health Mission (NDHM). The program, among other things, provides a framework for decentralized storage of patient owned data. Data standards and CDEs to be used for all health-related applications are well defined in the Meta Data & Data Standards documents from the Ministry of Health and Family Welfare. India does not have legislation protecting patient-data privacy such as the Health Insurance Portability and Accountability Act (HIPAA) in the United States, and the General Data Protection Rules (GDPR) in Europe. However, the Personal Data Protection Bill, which is not yet approved, allows patients control over their data and sets forth responsibilities for data users.

Given the current scenario in the country, patient-owned data collection efforts (registries) will benefit from the current trend of digitization and increased public awareness of health data. The authors recommend the pilot of a patient-owned rare disease registry in India and scale up in a gradual and dynamic manner allowing room to incorporate newer policies and laws that are underway.

AIMS OF THE FEASIBILITY STUDY

This initial phase of the project aims to conduct feasibility assessment for RARE-X in India and create a scoping document that will assess the opportunities to support patient-owned data collection in country. Steps of the study include:

1. Analysis of the landscape of rare diseases in India to understand current unmet needs from the perspective of patients and patient groups, and organizations, serving the rare disease community.

2. Analysis of the current regulatory and legal frameworks in India which are pertinent to patient data collection and sharing.

3. Interviews with stakeholders to assess interest and potential roadblocks for participation in a patient-owned data collection registry pilot:
   a. Feasibility assessment of an universal or commercial IRB for India pilots, and consider that all patients data collection needs to happen under an IRB approved protocol with proper informed consent and data privacy or security compliance equivalent to HIPAA and GDPR.
b. Review existing natural history data from PAGs in order to assess the quality and feasibility of importing or interfacing between various data.

c. Understand willingness of PAGs to participate in Natural History study, and to gather initial letters of commitment where feasible.

d. Identify potential investigators to develop India-specific protocols & guide the disease-specific data elements & collection methods. Gather initial letters of commitment where feasible.

e. Assess the extent of support available or gaps in operationalizing sharing of patient-owned data to advance R&D.

f. Understand regulatory support or roadblocks to consider for execution of this India pilot.

4. Develop strategy for education and outreach to patient advocacy groups on the importance of data, quality, privacy, and processes to address the significant lack of understanding of the need for and methods of collecting high quality data.

5. Discuss and define Common Data Elements (CDEs), data formats, standards in practice, recommendations on consistent and standardized collection of quantitative versus qualitative data and impact on data quality.

TIMELINE

<table>
<thead>
<tr>
<th>PROJECT TIMELINE</th>
<th>MONTH</th>
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<tbody>
<tr>
<td>Final Deliverable: Feasibility Assessment Report</td>
<td>1 2 3 4 5 6</td>
</tr>
<tr>
<td>Focus groups, interviews, data collection</td>
<td>**</td>
</tr>
<tr>
<td>Current state assessment</td>
<td>**</td>
</tr>
<tr>
<td>Desired future state definition</td>
<td>**</td>
</tr>
<tr>
<td>Identify gaps in technology, regulatory processes, training, and resources necessary to transition from current state to desired future state</td>
<td>**</td>
</tr>
<tr>
<td>Feasibility assessment / recommendation</td>
<td>**</td>
</tr>
<tr>
<td>2-year development roadmap with potential funding sources, legal and insurance frameworks.</td>
<td>**</td>
</tr>
</tbody>
</table>

Table 1: Gantt Chart showing the proposed timeline for the RARE-X India Pilot feasibility study by IndoUSrare. Month 1 = July 2020.

The projected timeline was estimated prior to the COVID-19 pandemic which has resulted in changes in strategy and slight time line shifts. The pandemic has impacted the team’s ability to interact with stakeholders, and all interviews were conducted online. It probably has also affected the response from candidate interviewees, due to altered workloads and schedules especially for physicians and regulatory representatives. The authors wish to also note that the project timeline was impacted by unavailability of KOLS as the last month overlapped with the holiday season.
1. **Literature Review:** An initial survey was conducted of existing information on the landscape of rare diseases, patient registries, and collection and sharing of health data. This includes a detailed review of scientific and regulatory literature.

2. **Rare Disease Stakeholders Mapping:** An initial list of candidate interviewees was made to include the following key classes of stakeholders. The team tapped into its existing network of rare disease stakeholders in India. Additional candidates were added to the list based on referrals provided by the initial set of interviewees.
   
i. **Patient Advocacy Groups (PAG):** The main class of stakeholders that represents a multitude of patients living with rare diseases spread across India. They are the voices representing their rare disease community and are key players in the rare disease scene in the country. As such, PAGs are reliable sources of information for current state assessment, and provide valuable insights.
   
ii. **Independent Patients and Disability Advocates:** Although less in number, there are also various individuals who are patients and advocates for rare diseases and disabilities and often not fully associated with a specific organization. They provide valuable inputs from the patient’s perspective.
   
iii. **Physicians:** The main interface between the patients and medical advancements in diagnosis and treatments. They are a key stakeholder in the rare disease landscape in a country like India where people from rural and lower socio-economic or educational backgrounds depend heavily on advice from their doctors.
   
iv. **Policy Professionals:** India’s rare disease policies are still evolving and personnel working at the frontline in this endeavor are good sources of information for current state assessment and can also provide inputs on implementation strategies.
   
v. **Industry:** This class of stakeholders includes Biopharma sponsors, Service Providers, Diagnostic Labs, and Contract Research Organizations (CRO). India has seen a growth in industry targeting the rare disease community, with the formation of a number of organizations that provide services or products for rare disease patients. These range from smaller startups as well as bigger companies with wider reach across the country and internationally. The successful functioning of these organizations depends on the availability of proper infrastructure and policies regarding rare diseases, and are hence important stakeholders in these discussions. While CROs do not specifically focus on rare diseases, they are the main drivers of clinical trials in the country and can thus provide valuable inputs for current state assessment.
3. **Design of Interview questionnaires:** Initial interview questionnaires were drafted for (i) Patients and PAGs, and (ii) Healthcare practitioners, Industry, and Researchers. Sample questionnaires are included in Annexures II and III.

4. **Interviews and Post interview follow ups:** Candidate interviewees were sent email invites which provided a brief introduction of a patient-owned rare disease registry project and requesting participation in a no-obligation discovery interview. Interviews were scheduled as per the candidate’s convenience and were conducted via web-based audio or video calls such as Zoom, Google Meet or WhatsApp. Zoom was the preferred mode for most interviews. Zoom interview sessions were recorded with consent/permission.

   All interviewees were sent follow up emails, thanking them for their participation and requesting introductions to possible interviewees in their network. Interviewees were also emailed to clarify any existing information and follow up with additional questions.

5. **Analysis of Data:** All interviews were recorded with permission, and transcribed into word documents. All answers were coded and then analyzed. A key highlights table was generated which captures aspects relevant to the main themes being analyzed.

A total of 102 individual email requests were sent to candidate interviewees. As of 28 December 2020, 28 candidates had accepted, 2 declined to participate in the study, and the remaining ones are yet to respond. We have made attempts to follow-up with non-responders, and those who had postponed interviews after acceptance, mindful of their privacy rights, after a month has elapsed.
RESULTS

LANDSCAPE OF RARE DISEASES IN INDIA

The first reported reference to rare diseases in India was in 1967. Since then, India has reported only 450 of the 7000 known rare diseases. These are all from the limited number of tertiary care centers in the country. Extrapolating from international data, there are an estimated 70 million people afflicted with rare diseases in India. Compounding factors include the diversity in the Indian populations with widespread practices of endogamy and consanguinity in many communities. Despite this large number, India faces numerous challenges related to management of rare diseases. One of the main challenges is the abysmally poor awareness about rare diseases. There are very few centers in the country with the infrastructure and technology needed to diagnose rare diseases, causing a majority of patients to go undiagnosed. Medical genetics is currently not an important aspect of the curriculum in medical college, resulting in a lack of awareness in the medical fraternity. This makes diagnosis a long-drawn process for even those patients who can afford and have access to genetic testing. Moreover, diagnosis is often just the first step of a tough journey for a vast majority of patients who cannot avail treatment because of the prohibitive price of the few available orphan drugs. This issue is often exacerbated because of the lack of health insurance for most people in the country. Furthermore, there is a paucity of research in rare diseases, which can be attributed to lack of infrastructure and funding for such work.

As is the case throughout the world, Patient Advocacy Groups (PAGs) in India have evolved out of the need to fill this gap in information, infrastructure, support, and policy. PAGs in India are mainly disease specific; but there are also a few umbrella organizations that cater to a wider range of rare disease patients. PAGs have made a significant impact on the rare disease landscape in the country. The organizations work to provide support to patients at the local and national levels. For instance, the umbrella organization, Organization for Rare Diseases India (ORDI) set up a national helpline for rare diseases. Patient groups also raise awareness through various campaign events in various cities and on television, radio, newspapers as well as social media outreach platforms. Special events are organized on dates such as the Rare Disease Day and awareness weeks and months for specific diseases. PAGs are also actively involved in facilitating and raising funds for research. Patient groups have also been actively involved in driving policy making in favor of rare diseases.

India has also seen an increase in the number of organizations offering medical products and services that have the potential to improve the lives of rare disease patients. These include genetic testing and diagnostic services, organizations involved in drug discovery and development for rare diseases, and companies that manufacture products that help in disease management such as motility devices and special diet foods. Despite the huge burden of rare diseases on India’s healthcare system and economy, the country is yet to have an official policy for rare diseases, including a definition for what is considered a rare disease.
However, these organizations face several issues such as lack of funding, unavailability of raw materials in a cheap and sustainable manner, and poor support from the government in the form of policies and incentives. Despite these hurdles, they have made significant strides and carved a niche for themselves in the Indian context. In one notable case, a parent led patient group, Dystrophy Annihilation Research Trust (DART) has funded and set up a research laboratory, which has now successfully developed an antisense oligo-based exon skipping treatment for Duchenne Muscular Dystrophy (DMD). This has been approved for pan India trials across multiple cities. This has led to calls for more patient driven research for rare diseases. A recent initiative has brought together clinicians and scientists to form a consortium of individuals to build a collaborative platform that aims to leverage the power of genomics and large scale analysis to understand and serve the needs of the rare disease community through systematic characterization and diagnosis of rare genetic diseases in India.

Despite the huge burden of rare diseases on India’s healthcare system and economy, the country is yet to have an official policy for rare diseases, including a definition for what is considered a rare disease. The much-awaited National Policy for Treatment of Rare Diseases (NPTRD) was announced in 2017. Formulated based on inputs of various stakeholders such as patient groups and researchers, the policy was a comprehensive document that detailed the public health concerns and economic burden of rare diseases, listing lack of epidemiological data, varying definitions and prevalence thresholds, lack of proper diagnosis, and limited research and development as key issues. It also outlined the key challenges in treatment of rare diseases. The policy called for implementing a holistic approach towards rare disease including multiple aspects such as awareness creation, education, prevention, treatment, and research. It also suggested ways in which the policy could be implemented by concerted action from multiple ministries and States. However, the policy was put in abeyance in 2018. After much uproar and protests from across the country, a revised National Policy for Rare Diseases was announced in January 2020, which drew a lot of flak from various patient groups, stating it is “far from reality” not taking into consideration the actual state of rare disease patients, and that it is a “mockery” of the plight of these patients.

There has been some progress such as the recent ruling by the national regulatory body for pharmaceuticals and medical devices, the Central Drugs Standard Control Organization (CDSCO), announced the New Drugs and Clinical Trials Rules, 2019 (NDCTR), which came into effect in 2019. This policy laid out a definition for an Orphan Drug, and provided key incentives for approval for these drugs such as waiver of requirement for local clinical trials for those orphan drugs which have already been approved and marketed in other countries, accelerated approval process and waiver of application fees.
UNDERSTANDING HEALTH INFORMATION REGULATIONS IN INDIA - IMPLICATIONS FOR GLOBAL PATIENT DATA SHARING

One of the highlights of the original well received NPTRD was the creation of a national registry for rare diseases. This led to the launch of the Indian Rare Disease Registry, designed and maintained by the Indian Council for Medical Research. Although this move was well appreciated, the registry is not yet functional as of 2020. Currently, India does not have a national level registry which provides accurate data on the distribution and prevalence of rare diseases in the country. The NPTRD also stressed the importance of multi-sectoral and cross border collaborations to tackle rare diseases. However, India does not yet have laws regulating collection, storage and sharing of health data.

The move to digitization is a welcome step, making accessible and affordable health care a reality for a large section of the population who have so far fallen through the cracks of India’s overburdened healthcare system. However, there have been several reports of the leak of health data of patients, sometimes of millions of records. A recent blog article looked into this situation and found that India’s hospitals store data in unsecured servers which can be accessed, and modified by even the minimally trained. This underscores the importance of ensuring that the country adopts strict standards and legislation that protects against such incidents. In this context, there are two main pieces of legislation worth noting:

1. **Personal Data Protection Bill 2019**
   
   The Digital Information Security in Healthcare information Act (DISHA) was proposed by the Ministry of Health and Family Welfare, Government of India (GoI) in order to “enforce privacy & security measures for electronic health data, and to regulate storage & exchange of Electronic Health Records”.

   It was later decided to incorporate the drafted legislation into the upcoming **Data Protection Framework on Digital Information Privacy, Security & Confidentiality’ Act**, currently known as the **Personal Data Protection Bill**, which is still not approved in parliament.

   The PDP Bill, introduced in Parliament in 2019, governs the processing of personal data by all data fiduciaries such as the government, and all companies within India and those that are based abroad. The bill defines what constitutes personal data as well as sensitive personal data. It provides for the protection of personal data of individuals, lays down a set of obligations from data fiduciaries, and establishes a Data Protection Authority for overseeing the same.

   The bill allows for sensitive personal data to be transferred outside India if there is explicit consent from the individual, and is compliant with other specific conditions. This data however, should continue to be stored in India. Additionally, there is a subset of personal data notified as critical personal data by the government that can only be processed in India. Of particular importance is the bill’s emphasis on the rights of a ‘data principal’, who are individuals whose personal data is being collected. This includes the right to provide or deny consent for disclosure of their data, the right to correct incomplete or inaccurate data, and the right to be forgotten. As such, they have the right to withdraw consent at any time.
2. **National Digital Health Mission (NDHM)**

The National Digital Health Mission (NDHM) was launched by the Prime Minister of India in his Independence Day address to the nation in 2020. This scheme was announced under the Ayushman Bharat Pradhan Mantri Jan Arogya Yojana, and builds on the National Digital Health Blueprint (NDHB), which aims to extend the existing National Health policy to ensure Universal Health Coverage for all Indians. Various digital systems will be employed for the purpose:

i. **Health ID:** Every Indian will get a Health ID that will be used to uniquely identify and authenticate the individual, and allows for sharing of their health records across multiple systems and stakeholders with their informed consent.

ii. **Digi Doctor:** The program aims to create a single repository of all doctors enrolled in nation which includes all their relevant details such as name, qualifications, specializations, registration number with State medical councils, years of experience, etc. would be an essential building block of the digital health infrastructure of the country. The registry is to be updated as doctors gain experience and expand skill sets.

iii. **Health Facility Registry (HFR):** The NDHM also calls for the creation of a single repository of all health facilities in the country, which is centrally maintained, and allows exchange of information from public and private health care facilities across the country, provide a secure common platform to the facilities to maintain all essential information. Facilities should be able to update their profiles periodically.

iv. **Electronic Medical Records (EMR):** will contain a digital version of a patient’s medical and treatment data from a single health facility.

v. **Personal Health Records (PHR):** is an app that electronically stores all health related information of the individual, in a manner that conforms with national interoperability standards. While the record can be built on information from multiple sources, full control regarding managing records rests with the patient. This can be integrated with other facilities such as telemedicine, e-pharmacy, and also used for creating a national health registry.

Key features of the NDHM include the following:

- Every Indian will get a Health ID that stores all medical records - This can be integrated with other facilities such as telemedicine, e-pharmacy, and also used for creating a national health registry.

- The NDHM will employ a federated architecture wherein regional, state and institution level platforms can be built to function in an independent yet interoperable manner. This decentralized approach allows data to be stored close to generating source.

- The system will expand upon the existing Digi Locker consent management framework to ensure that Health records can be accessed and shared by the patient using appropriate consent mechanisms. The patient will have complete control of the records.

- All Health care providers are expected to adopt systems that will allow them to Health Information Providers (HIPs) or Health Data Fiduciaries. HIPs will have a digital copy of both inpatient and outpatient health records. All HIPs will be enrolled in the NDHM health care infrastructure registry (HFR).
Any entity that intends to view health records of an individual patient is a Health Information User (HIU), and will have to request consent of the patient, without which they cannot access any data. HIUs will also be required to use software compliant with NDHM regulations.

The NDHM official website states that it ensures data security and privacy through its design. The NDHM also calls for a National Policy on Security of Health Systems and Privacy of Personal Health Records to be developed, in accordance with the PDP Bill 2019, which will guide development of all constituting systems that would handle personal health records.

The Health Data Management Policy of the NDHM was recently approved by the Government of India. The policy defines the minimum standard for data privacy to be followed, to ensure compliance with relevant and applicable laws, rules and regulations. However, since the policy is meant to act as a guideline in conjunction with existing laws, the success of the NDHM depends upon a proper law on data protection. The current lack of such a robust legal framework raises data privacy concerns.

**NDHM Sandbox:** The NDHM provides a platform that encourages entrepreneurs to be a part of the ecosystem, by providing a framework that allows technologies or products to be created in a contained environment in compliance with NDHM standards.

**DATA STANDARDS AND REGULATIONS IN INDIA**

The NDHB has recommended the adoption and use of several standards for health data. These include FHIR-R4, SNOMED-CT, LOINC, ICD10/11. Currently, there is extremely low adoption and implementation of standards among healthcare providers. The NDHM encourages gradual adoption of standards.

India’s Ministry of Health and Family Welfare has released the Health Domain Metadata and Data Standards (MDDS) document which describes the set of common data elements (CDEs) to be used for any new application developed in the Health domain in the country. However, since India is yet to have a formal, fully defined, and widely accepted policy for rare diseases, this list will need to be supplemented with CDEs listed elsewhere for rare diseases, including those from regulatory agencies such as the National Centre for Advancing Translational Sciences (NCATS), NIH in the US, and the EU RD platform.

Despite the lack of specificity to Rare Diseases, the MDDS is a comprehensive document that provides guidelines for the use of CDEs and processes. It identifies approximately 1000 data elements, grouped under 39 logical entities to ensure ease of use. The document also provides an extensive metadata directory containing the data type and size for each data element defined under MDDS allows for easy mapping and to ensure implementation of the recommended data standards. The document allows room for flexibility and the CDEs provided are meant to act as a superset from which program specific data elements can be defined.
IndoUSrare conducted a study to understand the rare disease landscape in the country with specific focus on patient data collection and sharing and patient registries. Interviews were conducted with different stakeholders to understand their interest in participation in a patient-owned health data registry and the feasibility and roadblocks for a platform that supports global sharing of patient owned health information.

Details of stakeholder mapping, and interview design have been provided in the Methodology section.

**OVERVIEW OF INTERVIEWS**

<table>
<thead>
<tr>
<th></th>
<th>Industry</th>
<th>PAG</th>
<th>Patient and Disability Advocate</th>
<th>Physician</th>
<th>Policy Professional</th>
<th>Policy Researcher</th>
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</tbody>
</table>

Fig. 1: Overview of the responses from the candidate interviewees. As of 31 December 2020, 102 email requests have been sent of which 32 candidates have been interviewed.

Thirty-two interviews were conducted. Annexure IV provides details of the interviewees and the organizations they represent. Four candidates postponed scheduled interviewees. All calls were recorded, and transcribed. Answers were coded and analyzed.

All interviewees were sent follow up emails, thanking them for participation and requesting contact information for possible interviewees in their network. This resulted in around 50 leads (PAGs, researchers, physicians, and government officials), some overlapping with the existing list of candidate interviewees.

The key themes that came out of the interviews have been described in the following sections.
**INTERVIEW WITH STAKEHOLDERS - OVERVIEW**

![Pie chart showing distribution across different categories of candidates interviewed. As of 31 December 2020, 32 stakeholders and KOLs have been interviewed.](image)

**Key Themes from Interviews**

1. **Ownership of a patient’s health information**

   When asked who they felt should be the owner of a patient’s health information, 63% of the interviewees were of the opinion that patients should be the owners of their own health information. These comprised all the types of stakeholders such as representatives of industry, patient advocates, physicians, as well as policy professionals. Among these, 5 felt that the Government and its institutions should be the custodian, while three interviewees felt that clinicians should be the custodian. Four interviewees (<15%) of stakeholders felt that the clinicians should be the owner of a patient’s health information. This too came from interviewees of different backgrounds. Two interviewees considered the government to be the rightful owners to the patient’s health information; one is a patient advocate, and the other a rare disease service provider. Two interviewees chose not to answer. Interviewees who did not feel the patient should be the owner justified their answer stating that in India, a vast majority of people especially those with a rural and less educated background, do not have the knowledge to be able to take health related decisions themselves and rely on their clinicians.
2. **Awareness in general public regarding newer technologies such as NGS**

Overall, the awareness in the general public was considered very poor. However, this has seen an improvement in recent years due to efforts of various groups as well as marketing campaigns from private diagnostic companies. Even though people are aware of the existing technology, it is often beyond their reach financially, which is a hindrance to these techniques being used widely. Additionally, there is a very small pool of specialists who can analyze and interpret the data fruitfully, which results in misinterpretation as well as sub optimal use of expensive tests.

3. **Assessment of existing patient registries in the country**

All the interviewees were of the opinion that India does not have good quality patient registries of national remit. Many of the hospitals, PAGs and individual research institutions may have their own database, but these are disparate and inaccessible. Many interviewees expressed hope in the Rare disease registry launched by ICMR. However, this registry is still not functional 3 years after launch, and does not cover all diseases. There have been few initiatives from various groups to fill this gap and a few pan India registries do exist. These include the Rare Disease Registry of India (RRDI) set up at the JK Lone SMS Hospital, Jaipur, the Birth Defects Registry of India (BDRI), and registries set up at national level institutes such as SGPGI, Lucknow, AIIMS- New Delhi, and CMC Vellore.
4. **Interest in participation in a patient-owned health data registry pilot**
   All interviewees expressed interest in a patient-owned rare disease registry initiative, and 30 showed interest in participation when a pilot is launched in the country. The two interviewees who declined to participate cited retirement and other personal issues as a reason. However, even among those who showed interest, a few qualified their answer by adding that as representatives of larger organizations, they require consent from member patients or institutional approval as applicable. Yet a few others felt the need for more information on the initiative and what was expected from their side before committing.

5. **Key Concerns for implementation of a patient-owned health data registry in India**
   The following emerged as key concerns regarding implementation of a patient-owned rare disease registry in India:
   
   a. **Data security and confidentiality**: Even when interviewees were reasonably convinced of the value of sharing data and maintaining registries to further research and drug discovery as well as to ensure representation in clinical trials, there were general concerns regarding data security. Specific questions asked were related to how data confidentiality would be ensured and how patient’s privacy would be protected.
   
   b. **Inhibition from Patients**: Indian society has a significant level of stigma associated with rare diseases and the phenotypic manifestations of many diseases, which stem from deep rooted cultural traditions and prevalent superstitions, as well as due to lack of awareness in the general public. This can result in an inhibition from the patient towards reporting or sharing data related to rare diseases.
   
   c. **Challenges in Implementation**: Another key concern was regarding the implementation of the project in the country. The interviewees felt that implementation of a project would have to overcome the myriad of bureaucratic and infrastructure related hurdles in the country.
d. **Cost to patient**: Interviewees, especially patient representatives, were worried about the cost to the patient including that of the medical tests that would be required as raw data.

e. **Logistics for data collection**: Questions were raised regarding the project implementation pipeline – will there be Central and State government involvement, what are the infrastructure requirements of the project, will the Indian medical system support the implementation of the project?

f. **Other concerns**: Other concerns raised included those regarding the legal, regulatory and jurisdiction aspects of data collection and global sharing, lack of time to enter large volumes of data.

6. **Suggestions for implementation of a patient-owned health data registry in India**

   The main suggestions offered for the successful implementation of a patient-owned rare disease registry pilot in the country were as follows:

   a. The consensus was that **collaborations between and concerted action from all stakeholders** is necessary for the successful implementation of a patient-owned rare disease registry project in India.

   b. While many interviewees believed that **government involvement is needed** to achieve maximum reach for a patient-owned health data registry project, because a major chunk of the population in India still depends on state run health schemes and insurance is largely unavailable. At the same time, however, they suggested that it is preferable to **work through the non-government sector** for better performance due to bureaucracy in government.

   c. Many interviewees stressed the importance of **including Awareness or Education campaigns** for patients and clinicians during the implementation phase, as lack of awareness is a key issue for rare diseases in India. Such campaigns would help them understand the benefits of a patient-owned health data registry program and the value of data sharing and registries in general.
d. Many interviewees felt it would be useful to include elements that would provide immediate or sustained benefits to the patient, such as the **inclusion of resources for networking, and information**. Such an initiative would help to attract people to use the platform and ensure continued use.

e. Clinicians interviewed felt that **minimizing paperwork** would help improve the entry of information as having numerous forms to fill and a multitude of data elements would discourage use of the platform.

f. Lack of time was one of the key concerns raised by clinicians, who felt that **help with manpower** for data collection and entry would be an encouraging step towards use of the platform.

g. One clinician interviewed suggested the **inclusion of imaging data** in the EHR data collection to improve the usability of data and allow for review at a later point of time.

h. One interviewee suggested the use of a **multi-level structured approach to data collection** to ensure maximum coverage. This would ensure that while detailed medical history is welcomed and encouraged, anyone with a minimal set of health information can access and add data.

### KEY INSIGHT #5

**Lack of Awareness**

“**The thought (of patient reported outcomes collection) has not yet arrived.**”

— Policy Professional,
National Healthcare Institute

**EDUCATION AND OUTREACH FOR PATIENTS**

Given that the patient registries are almost non-existent and there is a significant lack of understanding of the need for and methods of collecting high quality data, there is a need for concerted efforts towards education and outreach for patient advocacy groups with a focus on the importance of data quality, privacy, and processes.

Webinars are a key information dissemination and educational platform. The authors recommend webinars to disseminate knowledge about the importance of data quality, privacy and the methods to achieve these goals.
CONCLUSIONS AND RECOMMENDATIONS

The figure below summarizes our findings of the feasibility analysis conducted during July - December 2020, and lists out the different elements to be considered moving forward.

1. **SWOT Analysis**

   **STRENGTHS**
   - Novelty and uniqueness of the concept in India
   - A platform that will be cohesive with newly envisioned digital health in the country
   - Large number of interested participants
   - LOI secured from a premier institute and KOL
   - Existing avenues and pipeline to generate funding for the pilot
   - Credibility of team (RARE-X and IndoUSrare)
   - Successful registry programs in communicable diseases such as HIV

   **WEAKNESSES**
   - Lack of awareness regarding value of registries and awareness about newer genomic technologies
   - Patients’ expectation of immediate health benefit
   - Data security concerns among patient groups
   - Concerns regarding cost to patient
   - Social stigma associated with rare diseases

   **OPPORTUNITIES**
   - Education and outreach activities to raise awareness and interest among patients
   - New regulatory and legislative initiatives to digitize health in the country
   - Patient registries in rare diseases are largely untapped and no prominent competition (besides the Govt)
   - National Digital Health Mission (NDHM) provides Sandbox environment to develop tools
   - Significant opportunity for collaborations with Indian Govt and NGOs

   **THREATS**
   - Lack of infrastructure for efficient data collection
   - Lack of stable regulatory framework for data sharing
   - Lack of trust among stakeholders
   - Lack of unity in rare disease patient groups
   - Current strong nationalistic voices in India are calling for developing technologies in-house

   ![Fig. 4: SWOT analysis of the feasibility of a patient-owned health data registry, like RARE-X, in India.](#)

2. **Recommended Future Plans for a patient-owned health data registry in India**

   Based on our analysis, IndoUSrare recommends the introduction of a patient-owned health data registry, like RARE-X, in India in a phased manner. Even though India currently has various roadblocks as listed above for full-fledged implementation of a patient-owned registry allowing global data sharing, overall, India has been conducive to moving towards digitization, as evidenced by nationwide acceptance of digital payments in a fairly short period of time. Furthermore, the authors recommend leveraging the growing trend of digitizing healthcare and increasing awareness around data privacy and protection, and the individual’s rights to ownership of their own personal data.
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