

How Alexion Shortened Time to Rare Disease Diagnosis with a Master Dataset



The challenge of becoming data-driven

People living with rare diseases often have to wait for years for an accurate diagnosis and many are never diagnosed at all. Even after diagnosis, only 5% of the 7,000 plus known rare diseases have approved therapies. Data plays a key role in diagnosing rare diseases and in the research behind their therapies – in finding insights, experimenting, and discovering new drugs.

Alexion, an AstraZeneca subsidiary, is a global biopharmaceutical company that develops drugs to treat rare disorders. Its goal is to leverage the best available medical data for rare diseases and clinical insights to accelerate the diagnosis of rare conditions.

The Impact of Lacking a Central, Trusted Source of Rare Disease Definitions

While there is more medical data available than ever before, much of it remains untapped, disorganized, and unusable. For instance, there is no single authority providing a comprehensive list of rare disease definitions. In fact, there isn't even a consensus on the number of rare diseases that exist.

Company

Alexion

Alexion is a global biopharmaceutical company focused on developing life-changing therapies for people living with rare disorders.

Pharma Analytics

Drug Discovery

Rare Disease Diagnosis

Depending on the source consulted, the answer varies between 7,000 and 9,000 rare conditions. Meanwhile, Alexion's dataset contains over 12,000 conditions with 7,230 from Orphanet – and many more are in the process of being defined.

Without a trusted and accurate source of rare disease definitions to refer to, it is difficult for physicians to correctly and quickly identify a diagnosis. Most rare diseases lead to considerable disability and early mortality, so a rapid and accurate diagnosis is crucial. Even after receiving a diagnosis, close to 90% of people with a rare disease do not have an approved treatment or therapy. The grim reality is that, of the thousands of rare conditions, only 161 have approved drugs available. The lack of a recognized inventory of rare disease definitions leads to under-representation in healthcare planning and resource allocation and prevents clinical research from being carried out.

What's more, the lack of a rare disease master dataset also has an impact on medical insurance coverage. The healthcare system in the United States uses ICD-10 codes for billing. There are barely 128 ICD-10 codes for over 7,000 rare conditions, which means that only a minuscule number of rare diseases are recognized by insurance providers and get coverage.

Ultimately, a rare disease definition is the backbone to which all clinically relevant information is mapped, and a lack of it leads to delays in every aspect of disease management from drug discovery, clinical trial design, and drug approval to diagnosis, treatment plan, and insurance coverage. Alexion wanted to address these challenges by building a rare disease data master to shorten the diagnostic journey and time to initiate treatment for those living with rare conditions.



Ingesting Constantly Changing Medical Data

Alexion adopted KNIME as its core analytics tool to accelerate the rare disease diagnostic odyssey. With KNIME, they were able to easily ingest data from countless biomedical knowledge bases such as **Orphanet**, **PubMed**, **DrugBank**, **Reactome**, and **GTex** and get it into the right shape. Before KNIME, if Orphanet or any other data source changed its data format, the team at Alexion would end up spending up to three weeks modifying their Java code, indices, and APIs to accommodate the new format. With KNIME, they were able to seamlessly integrate and transform the data in one uniform, visual environment in no time, regardless of any changes within the data source.

Once the transformed data was loaded, KNIME enabled Alexion to build all-inclusive, master datasets such as the disease master, drug master, gene master, and trial master that helped close major gaps in rare disease information.

Accurate and Actionable Rare Disease Definition Dataset

This data was sent for validation to clinicians and subject matter experts through an interactive dashboard on a KNIME-built data app. These stakeholders - typically PhDs with over 20 years of experience - often insisted on seeing the data lineage and the business logic that was followed for data mapping before they provided feedback. KNIME's ability to automatically document each step of the data wrangling process enabled Alexion to provide stakeholders the complete visibility they needed into how data pipelines transitioned from raw data all the way to analysis. The feedback from clinicians has led to the creation of a master dataset that is 80% actionable and its quality continues to improve with time.



Shortened Rare Disease Diagnostic Journey

Today, this rare disease data master serves as an accurate and ready reference tool for physicians to deliver a precise diagnosis quickly. In some cases, the diagnostic time has been brought down to a few hours from several years by matching against the rare disease master.

The rare disease data master also equips physicians with the information they need to provide early and effective treatment for patients after diagnosis. It reduces dependence on unreliable Google searches and addresses the clinical knowledge gap.

Additionally, the rare disease data master forms the center of a flywheel of patient-centric communities around genetics, symptoms, and treatments that work, and it holds the potential to fast-track discovery and approval of new therapies.

Improved Rare Disease Recognition and Medical Coverage

A single, trusted inventory of rare disease data bolsters the chances of higher rare disease representation in healthcare coding systems such as ICD. This would lead to increased interest in clinical research from the healthcare community as well as the much-needed recognition of rare conditions by insurance providers.

Why KNIME

KNIME's open-source nature was a key factor in it becoming the tool of choice for Alexion. Multiple research institutes that Alexion partners with, were able to start using KNIME without any barriers on scaling. The ability to share complex data with clinicians and subject matter experts through an easy-to-use, interactive data app coupled with the ability to track data lineage was invaluable for Alexion. With KNIME, Alexion and its partners were able to work on the same analytical pipeline while executing it independently on their own datasets. In other words, it helped them bring the analytics to the data and not the other way round.