Category

Best Startup

Product/Solution Name

RDK Rare Disease Knowledge

Date of Approval

N/A

Indications

PRODUCT / SOLUTION

Company Name
As We Know™ is a French company whose purpose is to share medical and scientific knowledge by developing digital tools for healthcare professionals and patients to facilitate disease management

Company information
As We Know™ is a mission-driven organization characterized by social or environmental objectives (French Pact law of May 22, 2019) and is committed to:
· Respecting its purpose and missions
· Donating 1% of its revenue to patient associations
· Appointing a mission referent.

Product/Solution Name
RDK Rare disease Knowledge ™
· RDK Rare disease Knowledge™ is an application (mobile and web) that allows healthcare professionals to access the latest available knowledge on rare diseases and expert centers to reduce diagnostic errancy for patients*.
RDK is codeveloped with Orphanet (Inserm)

Date of Approval
French Health Authority : ANSM registration as SaMD class 1 January 19, 2023 (Software as Medical Device) .CE marking as of July 12, 2022.

Sub-Categories (Digital Health /Medtech / Biotech)
Software as Medical Device

Therapeutic Categories

Rare Diseases

Attached Files:

- AWK Submit your innovation_Prix Galien USA_Startup.pdf

Background information and need for solution/product

Background information

• Diseases are said to be rare when they affect one person in 2,000, i.e. more than 3 million French people and at least 30 million European citizens.
• Today, 7000 rare diseases are identified and 80% are genetic
• A patient with a rare disease must endure an average of 4 to 5 years of diagnostic errancy
• Rare diseases can be hidden behind fairly common symptoms. This can lead to misdiagnosis and delays in care.
• Improving information and access to information is one of the major challenges of the rare disease plans. Organizing diagnostic assistance starting from the general practitioner and relying on the rapid identification of a referral, on the use of networks and new information technologies is another equally important issue.

Need for solution/product

Faced with 7000 rare diseases and thousands of symptoms (more than 13000 that characterize the rare diseases) it is necessary to find a unique solution accessible to all doctors in 1st line with the patients to :
• access to the last available knowledge
• help the identification of diseases from common symptoms and to direct patients
• refer patients to the appropriate expert centers

Product General Information

• RDK Rare disease Knowledge™ is an application (mobile and web) that allows healthcare professionals to access the latest available knowledge on rare diseases and expert centers to reduce diagnostic errancy for patients*.
• The application is free for doctors in order to guarantee a large diffusion and is financed by industrial sponsors, institutions or any actor of the health sector wishing to show his commitment in the fight against rare diseases.
- RDK is A unique public-private partnership based on Orphanet's expertise, Tekkare® technology, As We Know®' societal commitment and the support of sponsors
• As We Know™ was created specifically as a mission-driven organization (“Entreprise à mission , loi pacte 22 mai 2019”) to distribute RDK Rare disease Knowledge™

History of the development of the solution/product

RDK has been developed in partnership with Orphanet (Inserm) and Tekkare (health tech).
A complementary technical and scientific team (Orphanet) together with the aim of providing a solution to reduce diagnostic errancy, with total commitment on both sides.
The project leader, with dual French and American nationality, and the international profile of the teams, enabled us to design an application that could be natively multilingual.

• March 2021 : RDK development began
• June 2022 : The first proof of concept was presented
• July 2022, submission to ANSM (French health authority) of the registration file as SaMD class 1
• January 2023 : Confirmation of RDK's compliance registration by the health authorities ANSM
• June 2023 : launch with physicians

RDK main features

· Screening tool : Based on a combination of symptoms observed, the screening tool guides the practitioner towards a list of rare diseases.
o Results are classified by relevance according to symptom frequency and diagnostic criteria.
· Exploration of the 7000 rare diseases with the latest knowledge updated every month
o Information & data
 Definition, synonyms, heredity, age of onset
 Epidemiology
 Etiology, diagnostic methods, differential diagnosis
 Genetic councelling
 Management of treatment
 Prognosis
 Genes
o Case report and review of the disease updated every day (based on Pubmed)
o Clinical trials updated every day (based on clinicaltrial.gov)
· Directory of expert centers classified by rare disease
for each center, the name of the rare disease network, the name and address of the hospital, the name of the department, its email and telephone number

· Data sources used in RDK
o Orphanet
o HPO
o Pubmed
o Clinicaltrial.gov

· Data collection and publication
o Privacy : No patient or physician data is collected to ensure complete confidentiality and compliance
o Usage statistics by physicians are monitored to track the real-life impact on the daily practice of doctors and patients alike
o Statistics on the most sought-after symptoms and rare diseases will be used for publications and to improve the application using machine learning techniques

The doctor’s journey in the RDK application
the doctor has a patient with a difficulty of diagnosis, he suspects a rare disease but which among the 7000 rare diseases?
The doctor launches RDK on his cell phone or computer, he enters the symptoms of his patient and discovers which rare diseases have these symptoms with a relevance score. He can then consult all the updated knowledge on this disease and find the expert centers. Directly in the application he can contact the nearest expert center by email or phone to refer his patient. (cf pdf RDK doctor Journey)

Future development for RDK
New versions : a road map has been defined with Orphanet for V2 and V3, which will include innovative new functions (confidential information).

International development :
· The RDK application is already multilingual in English and French, and other languages will be added as the international rollout progresses.

· the main part of the application is identical in all countries (symptoms and rare diseases). For local deployment, all we need to do is adapt the application to the country's expert centers.
· Our focus is to launch first in
o USA : We would like to set up a unique tripartite partnership with Orphanet and NIH (Gard).
o Europe : Orphanet is already part of the European organization for rare diseases (EJPRD)

Financing:
the application is financed by sponsorship from all those wishing to commit to the fight against rare diseases: life science company, institutions, foundations, associations, etc.
The first two sponsors are Pfizer and Amylyx. We expect more soon.

Attached Files:
- AWK_RDK_Deck_journey.pdf

Why this solution/product is innovative, the broad implications for future research, and/or how it will improve the human condition

Why this solution/product is innovative
RDK will be the first mobile (and web) application available to address the problem of diagnosis errancy for all 7000 rare diseases in coordination with the main French public actor involved in the fight against rare diseases Orphanet™ (Inserm), the world reference.

By combining data processing technologies, application development and validated scientific data, RDK offers a daily tool for doctors to help them detect rare diseases and improve patient care by directing them more quickly to the appropriate expert centers.

The broad implications for future research
The application will then deployed very quickly throughout Europe and the United States.
the data collected in real life will allow to understand the symptoms, diagnostic paths, and the rare diseases most sought after by doctors in the first line.
They will be the subject of research and publications and will help to improve the search engines and the application algorithms (machine learning).

how it will improve the human condition
By enabling earlier detection of diseases and directing patients to expert centers, the application will reduce the physical and psychological damage caused by patient errancy.

Please provide appropriate references (ie Pubmed links)

· ERRADIAG https://www.alliance-maladies-rares.org/wp-content/uploads/2020/05/Erradiag-l-errance-diagnostic-dans-les-maladies-rares1.pdf
· L'errance diagnostique : le point de vue des maladies https://www.medecinesciences.org/en/articles/medsci/full_html/2018/05/medsci180152s/medsci180152s.html

· L'errance diagnostique dans les maladies rares 2018 https://dumas.ccsd.cnrs.fr/dumas-01932874

· The health and life path of rare disease patients: results of the 2015 French barometer

https://www.dovepress.com/the-health-and-life-path-of-rare-disease-patients-results-of-the-2015--peer-reviewed-fulltext-article-PROM

· Maladies rares : les médecins généralistes contribuent à réduire l'errance diagnostique 2022 https://www.vidal.fr/actualites/29242-maladies-rares-les-medecins-generalistes-contribuent-a-reduire-l-errance-diagnostique.html

Attached Files:

- Letter Prix Galien.pdf