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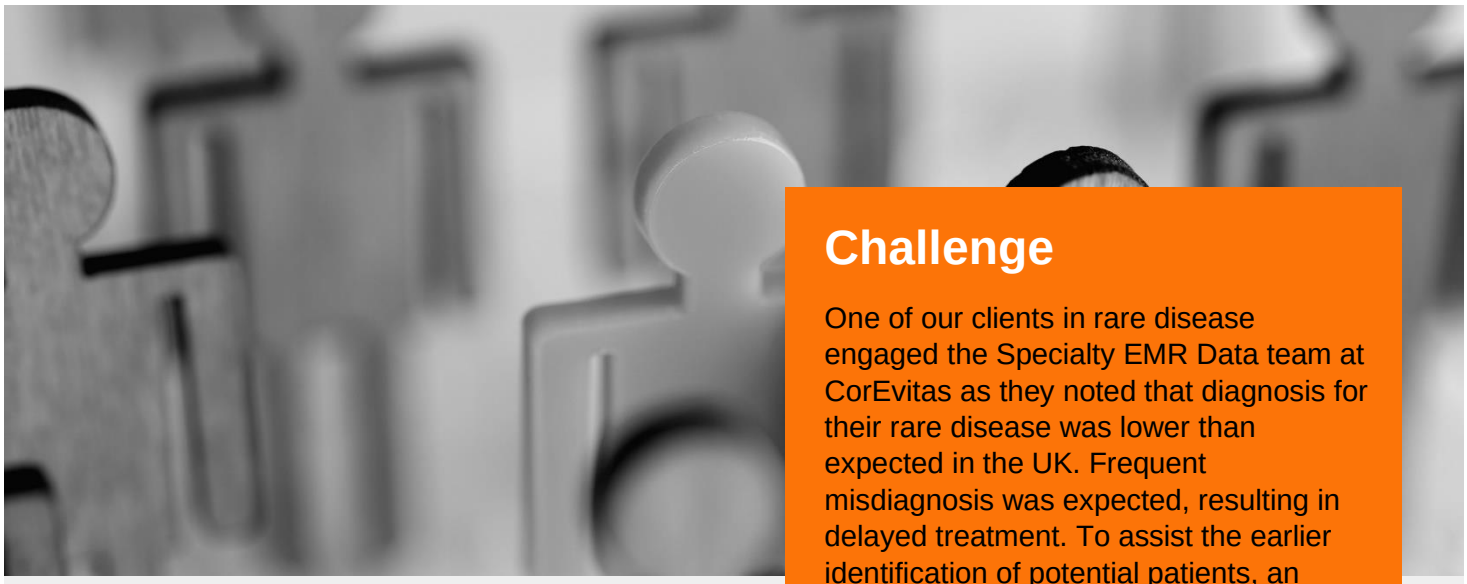
Case study: Supporting earlier access to diagnosis in rare disease



Challenge

One of our clients in rare disease engaged the Specialty EMR Data team at CorEvitas as they noted that diagnosis for their rare disease was lower than expected in the UK. Frequent misdiagnosis was expected, resulting in delayed treatment. To assist the earlier identification of potential patients, an England-wide cohort of all patients diagnosed with the condition was identified and analysed to discover the current pre- and post-diagnosis pathway. Patients with the condition were characterised and relevant inclusion and exclusion criteria formulated to identify the size of a potential 'screening population' who meet the characteristics of a patient with the condition but are yet to be diagnosed.

Background

There are often diagnostic challenges in rare diseases. These challenges occur due to the diversity of symptoms, which are often non-specific. Due to the rarity of the condition, clinicians will also not commonly see patients who present with the disease in question. Such rare diseases can often go un-diagnosed or misdiagnosed for several years, leading to poorer outcomes for patients and increased burden to the healthcare system.

Solution

Using ICD10 diagnosis codes within the Hospital Episode Statistics (HES) dataset and separately SNOMED, plus the ICD10 diagnosis codes within the Clinical Practice Research Datalink (CPRD) and HES linked dataset, patients with a recorded diagnosis of the condition were identified.

Pre-diagnosis symptoms, consultations and medications were described in addition to secondary care interactions. Kaplan-Meier curves were produced to represent the time from first presentation of relevant symptoms to the time of diagnosis.

In combination with clinical consultation, the output from this analysis was utilised to formulate an inclusion and exclusion criterion for patients who potentially had the condition of focus but were yet to be diagnosed. This was then applied to the wider datasets to establish regional variation in number of potential patients.

Outcome

The opportunity for early diagnosis was highlighted and the results were submitted for peer-review publication in a medical journal. The diagnosis of the condition has seen increased threefold. This benefitted our client's product commercialization, but more importantly it enabled more patients to access their life-changing treatment sooner. In partnership with our client, our team won the PM society innovation award for this project.