

PROJECT OUTCOME SUMMARY

Feasibility study to find improved pathways to diagnosis of mucopolysaccharidoses

- ▷ MedicinesInsight is the leading large scale general practice data set established to support quality improvement in Australian primary care and post-market surveillance of medicines, it is developed and managed by NPS MedicineWise. MedicinesInsight extracts longitudinal, de-identified, whole-of-practice data from the clinical information system (CIS) of consenting general practices to connect patient conditions with treatments and their outcomes.
- ▷ BioMarin and Sanofi Genzyme co-sponsored VentureWise to conduct this project exploring the feasibility of using MedicinesInsight general practice data to improve the understanding of diagnostic markers for mucopolysaccharidoses (MPS) and to assist with early diagnosis. NPS MedicineWise was commissioned by VentureWise to analyse MedicinesInsight data and report on their findings and recommendations. NPS MedicineWise undertook this project with complete independence from BioMarin and Sanofi Genzyme, and BioMarin and Sanofi Genzyme had no influence or involvement in the project.
- ▷ Information from the feasibility study may be used as the basis for further analysis of MedicinesInsight data to determine what patterns of presentation may facilitate an earlier diagnosis of MPS, and also educational activities for GPs that may help connect patients to an earlier diagnosis and best practice management. As this was a feasibility study to understand what data was available within MedicinesInsight about MPS, the project team did not engage with consumers about this project. However, any future, more in-depth research/education project will engage with consumers, clinicians and other relevant stakeholders.
- ▷ Approval from the MedicinesInsight Data Governance Committee was obtained on 22 February 2017 to explore the MedicinesInsight data, identifying patients with a diagnosed MPS and analysing their documented diagnoses and treatments.
- ▷ Data of patients with a recorded diagnosis of MPS were examined to determine the possibility of finding recorded conditions that could assist with the identification of undiagnosed patients with MPS. Patients with MPS were identified using selected search strings. Records for individuals with a MPS diagnosis were examined for information including demographic data (patient gender, age group and state of general practitioner (GP) practice), all reasons for encounter, all diagnoses recorded and reason for prescription.
- ▷ A MPS prevalence of four in a million patients, a total of 41 cases, was identified in MedicinesInsight data as of February 2017. Of those patients with a recorded MPS diagnosis, 49% were aged under 20 years and 68% were male.
- ▷ About a third of the MPS patients had respiratory tract infections and ear problems, such as grommets, recorded in MedicinesInsight. About a quarter had gastrointestinal, musculoskeletal, psychological and skin problems and 12% had a hernia recorded. Combinations of conditions in patients with MPS found 17% with both ear problems (such as grommets) and gastrointestinal problems and 15% with both ear problems and respiratory tract infections (RTIs). As these are common problems in the general population, they do not assist with the identification of MPS patients. A high rate of grommets (63%) was found in children with MPS aged under 10 years. Significantly, only four of the 41 patients had any diagnoses recorded prior to the date of the first record of MPS and so there was minimal information to determine pathways to diagnosis or conditions known to precede an MPS diagnosis.

- ▷ MedicineInsight data was determined not to be useful for identifying diagnostic markers or combinations of conditions to base an algorithm for identifying potential undiagnosed cases of MPS. This was due to the identification of a wide range of comorbidities in general with limited commonality. The most common multiple comorbidities are common health problems. There was limited information in the database recorded prior to an MPS diagnosis.
- ▷ Patient journey mapping may be a suitable way to identify health professionals who could benefit from education on MPS and perhaps expedite referral and diagnosis.
- ▷ Education targeting GPs and utilising paediatrician expertise could include activities on the topic of 'red flags' for paediatric referral, or could cover rare diseases as a group. MPS is likely to be considered too rare to be of real interest to GPs as a stand-alone topic.

Suggested citation: MedicineInsight. Project Outcome Summary – Feasibility study to find improved pathways to diagnosis of mucopolysaccharidoses. Sydney: NPS MedicineWise, July 2018.

More information: for more information about this project, access to the full report or MedicineInsight enquiries, please visit <https://www.nps.org.au/medicine-insight> or email DataGovernance@nps.org.au