A POPULATION-BASED COHORT OF GAUCHER DISEASE PATIENTS IDENTIFIED USING EHR DATA

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INTRODUCTION

- Gaucher disease (GD) is an autosomal recessive disorder caused by a mutation in the glucocerebrosidase gene. Over 90% of patients have the type 1 non-neuropathic phenotype of GD. The incidence of type 1 GD worldwide is low, at 1 in every 100,000 live births, but is exceptionally high among people of Ashkenazi Jewish descent, at 1 in every 850 live births.1
- Patients with this orphan disease exhibit a broad range of symptoms and disease severity that manifest at all ages. GD-related complications and outcomes include hepatosplenomegaly, splenomegaly, bone events and cytopenia.

OBJECTIVES

- To characterize a population-based cohort of patients with GD relative to the general population.
- To describe sociodemographic and clinical differences by disease severity, as defined by the use of enzyme replacement therapy (ERT).

METHODS

Data source

- A cross-sectional study of patients with GD was conducted as of December 31, 2014 using the Clalit Health Services (CHS) electronic health record (EHR) database.
- CHS is the largest health care maintenance organisation in Israel, having >4.2 million insured residents and providing care to >75% of the Israeli population.

Study population

- Included were GD cases as of June 30, 2014, identified using a combination of the following (Figure 1):
  - A physician’s diagnosis in the patient’s permanent record or at least 83,084 (0.5%) 174 468 270 549,941 (9.4%) (93.6%) (20.4%) 1 3.6% (14.1%)
  - The prevalence of GD ranged from 5.3 per 100,000 for members ≤34 years of age to 20.2 per 100,000 for those ≥35 years of age. The age-standardised prevalence (according to the Israel population) is 11.6 per 100,000.
  - The majority of GD cases were in ages of GD = 91.6% of Clalit (68.3%), female (GD = 54.0% Clalit = 51.1%), primarily Jewish (GD = 93% Clalit 73.4%) and of higher income-status relative to the general Clalit population (GD = 34.8% vs Clalit = 19.0%) (Table 1).
  - The prevalence of overweight/obesity (among those with documentation) were 35.5% for GD cases and 46.6% among all Clalit members. 63.5% of patients with GD had a Charlson Comorbidity Index (CCI) ≥1, compared with 30.4% in the general population (Table 1).
  - The majority of GD cases had a history of anemia (69.6%) or thrombocytopenia (50.2%). 40% had a history of a bone event and 22.2% had a history of cancer (Table 2).

Analyses

- Analyses were performed descriptively to characterize the cases’ demographics, medical history, clinical characteristics and disease management.
- Clalit members as of December 31, 2014, excluding patients with GD, were used for the comparison of socio-demographic characteristics.
- Disease prevalence was calculated using the Clalit population and adjusted to the Israeli population of 2014.
- Chi-square testing was used to assess distributional differences between groups.
- IRB approval was obtained for all analyses.

RESULTS

GD population – general

- The sources of the EHR data were used to identify the 500 prevalent GD cases (Figure 1).

- The prevalence of GD ranged from 5.3 per 100,000 for members ≤34 years of age to 20.2 per 100,000 for those ≥35 years of age. The age-standardised prevalence (according to the Israel population) is 11.6 per 100,000.

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- The majority of GD cases had a history of anemia (69.6%) or thrombocytopenia (50.2%). 40% had a history of a bone event and 22.2% had a history of cancer (Table 2).

- Clinical characteristics, such as body mass index and smoking, were similar between ERT use groups, with the exception of CCI ≥1 (ERT+ = 79.4% vs ERT- = 52.3%, p<0.001) (Figure 2).

- Crude rates of disease complications such as anemia, thrombocytopenia and hepatomegaly were more common in ERT+ vs ERT- patients (p<0.002) (Figure 3). This was expected, as those with disease manifestations were more likely to be treated.

CONCLUSIONS

- This is the first large-scale socio-economic and clinical data on a large population-based GD cohort using a comprehensive real-time EHR database.
- The prevalence of GD ranged from 5.3 to 20.2 per 100,000 for patients under and over 35 years of age, respectively.
- The GD population differed socio-demographically and clinically from that of the general Clalit population.
- Statistically significant differences between those who had and had not initiated ERT treatment were observed for disease-related complications, but not for non-disease-related complications.
- By 2014, 41.2% of all patients with GD in the Clalit EHR database had been treated with ERT.
- Monitoring such a cohort is important to understanding disease burden and outcomes in a real-world population.

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DISCLOSURES

Flaks-Manov, A Benis, H Gabay and M Leventer-Roberts are employees of Micromedex Research Applications LTD, a subsidiary of Clalit Health Services. DH Jaffe is an employee of Kantar Health and M DiBonaventura was an employee of Kantar Health when the research was conducted. A Joseph is an employee of Shire International GmbH.

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