Global epidemiology of Gaucher disease: an updated systematic review and meta-analysis

Meimei Wang
China National Center for Food Safety Risk Assessment
Fengqin Li
China National center for food safety risk assessment
Jing Zhang
Beijing Friendship Hospital
Cheng Lu
Beijing Hongjian Medical device Company
Yan Meng
Chinese PLA General Hospital
Yingxue Ding
Beijing Friendship Hospital
Weijing Kong (kwj86213@126.com)
Beijing Friendship Hospital

Research

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Abstract

Background

Gaucher disease, an autosomal recessive lysosomal storage disorder, is characterized by progressive lysosomal storage of glucocerebroside in macrophages predominantly in bone, bone marrow, liver, and spleen. Meta-analysis of global Gaucher disease epidemiology was not available prior to this study.

Methods

To provide a systematic review and meta-analysis of birth prevalence and prevalence of Gaucher disease in multiple countries. MEDLINE and EMBASE databases were searched for original research articles on the epidemiology of Gaucher disease from inception until July 21, 2021. Meta-analysis, adopting a random effects logistic model, was performed to estimate birth prevalence and prevalence of Gaucher disease.

Results

Eighteen studies that were screened out of 1874 records were included for data extraction. The studies that fulfilled the criteria for inclusion involved 15 areas/countries. The global birth prevalence of Gaucher disease was 1.5 cases (95% CI: 1.0-2.0) per 100,000 live births. The global prevalence of Gaucher disease was 0.9 cases (95% CI: 0.7-1.1) per 100,000 inhabitants.

Conclusions

To our knowledge, this is the first comprehensive systematic review that presented quantitative data by evaluating global epidemiology of Gaucher disease. Quantitative data of global epidemiology of Gaucher disease could be the fundamental to evaluate the global efforts that improve many factors, including diagnostic technology and data collection, which affect global epidemiology of Gaucher disease.

Background

Gaucher disease (GD), an autosomal recessive lysosomal storage disorder, is characterized by progressive lysosomal storage of glucocerebroside in macrophages predominantly in bone, bone marrow, liver, and spleen (1). There are three subtypes of GD, which are mostly caused by pathogenic mutation of gene for glucocerebrosidase (2). Very rarely, deficiency in the GCase activator, saposin C, could cause GD (3).

Based on such pathogenic mechanism, there are two specific ways to treat GD: 1) recovery of enzyme activity, such as enzyme replacement therapy; 2) reduction of accumulation of glucocerebroside in lysosome, such as substrate reduction therapy (4). For now, there are several treatments for GD that have been approved, such as Cerezyme® and Vpriv® (1).

There was only one comprehensive review about GD epidemiology (5). Nalysnyk et al. (2017) (5) presented that standardized birth prevalence of GD in the general population varied from 0.39 cases to 5.80 cases per 100,000 live births, and prevalence ranged from 0.70 cases to 1.75 cases per 100,000 inhabitants, respectively. This study aims to show more precise results by updating previous systematic review and presenting quantitative epidemiological data of GD.

Methods

Literature search strategy

Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) statement was the guideline for this systematic review and meta-analysis (6). The complete checklist could be found in **Additional file 1**. The study strategy adopted to identify studies was as follows:

EMBASE and MEDLINE were searched by terms ("incidence", "prevalence", "epidemiolog*" and Gaucher disease") from inception until July 21, 2021. Endnote X7 was used to manage citations. Detailed literature search strategy for different databases was provided in **Additional file 2**.

Inclusion and Exclusion Criteria

Studies fulfilled all of the following criteria were selected: 1) the case collection was based on field survey; 2) the study was based on population samples rather than volunteers; 3) the study had definite numerator (number of patients) and denominator (number of live births or inhabitants).

Studies fulfilled any of the following criteria were excluded: 1) study without information available for meta-analysis was excluded; 2) conference abstract was excluded; 3) study that focused on one specific population from one area/country was excluded.

Quality of Studies

Quality of studies was assessed independently by two reviewers (MM W and FQ L) based on a checklist specifically for observational studies concerning rare diseases epidemiology, which was adapted from STrengthening the Reporting of OBservational studies in Epidemiology (STROBE) (7, 8). Details about this checklist were shown in **Additional file 3.**

Data analysis

Data extraction was operated independently by two reviewers (MM W and FQ L). For each included study, birth prevalence/prevalence per 10,000 individuals was considered as the primary outcome for meta-analysis. Stata/SE version 15.1 software (StataCorp LP, College Station, Texas) was used to conduct statistical analysis. Heterogeneity of epidemiological estimates, along with its derived measure of inconsistency (I^2), was assessed by Cochran's Q test (9). When $P < 0.1$ for the Q test or $I^2 > 50\%$, the signs for

substantial heterogeneity, were received, random effects model was used; otherwise, fixed effects model was performed. In addition, funnel plot was used to describe potential publication bias.

Results

Figure 1 showed the process of identifying eligible epidemiological studies. Eighteen studies, all of which met inclusion criteria and were not excluded by exclusion criteria, were selected and then subjected to quality assessment. Five (28%) studies were rated as high quality, 9 (50%) studies were considered to be of medium quality, and 4 (22%) studies were assessed as low quality (Table 1). Details about the quality of each included study were reported in **Additional file 3**.

Table 1
Quality reporting of included studies.

Author, Year of publication	1. Was there an adequate description of study design and setting?	2. Was there an adequate description of eligibility criteria?	3. Is the study population representative of the target population?	4. Is there an adequate description of outcomes?	5. Is there an adequate description of the study participants?	Overall assessment
(Meikle et al., 1999) (20)	Yes	Unclear	Yes	Yes	Yes	Medium
(Poorthuis et al., 1999) (30)	Yes	Yes	Yes	Yes	Yes	High
(Applegarth et al., 2000) (21)	Unclear	Unclear	Yes	Yes	NO	Low
(Dionisi-Vici et al., 2002) (22)	Yes	Yes	Yes	Yes	Yes	High
(Asuman Özkara and Topcu, 2004)(23)	Yes	Unclear	Yes	Yes	Yes	Medium
(Revest et al., 2009) (34)	Yes	Unclear	Unclear	Unclear	Yes	Low
(Poupetova et al., 2010) (31)	Yes	Yes	Yes	Yes	Yes	High
(Giraldo et al., 2012) (35)	Yes	Yes	Yes	Yes	Yes	High
(Mechtler et al., 2012) (24)	Yes	Unclear	Yes	Yes	Yes	Medium
(Stirnemann et al., 2012) (25)	Yes	Yes	Yes	Yes	Yes	High
(Liao et al., 2014)(36)	Unclear	Unclear	Yes	Yes	Yes	Medium
(Hult et al., 2014)(26)	Yes	Unclear	Unclear	Yes	Yes	Medium

More details of quality assessment could be found in **Additional file 3**.

Author, Year of publication	1. Was there an adequate description of study design and setting?	2. Was there an adequate description of eligibility criteria?	3. Is the study population representative of the target population?	4. Is there an adequate description of outcomes?	5. Is there an adequate description of the study participants?	Overall assessment
(Hopkins et al., 2015) (27)	Yes	Unclear	Unclear	Yes	Yes	Medium
(Burton et al., 2017) (37)	Unclear	Unclear	Unclear	Yes	Yes	Low
(Kang et al., 2017)(38)	Unclear	Unclear	Unclear	Unclear	Yes	Low
(Hopkins et al., 2018) (39)	Yes	Unclear	Unclear	Yes	Yes	Medium
(Burlina et al., 2018) (40)	Yes	Unclear	Unclear	Yes	Yes	Medium
(Chien et al., 2020)(28)	Yes	Unclear	No	Yes	Yes	Medium
More details of quality assessment could be found in **Additional file 3**.						

The studies that fulfilled the criteria for inclusion involved 15 areas/countries. Table 2 showed characteristics of each study. As shown in the table, 10 (56%), 4 (22%), 3 (17%) and 1 (5%) studies were from Europe, North America, Asia and Oceania, respectively. Primary results showed $P < 0.1$ for the Q test or $I^2 > 50\%$, so random effects model was used. Variables from outcome measures were pooled using DerSimonian and Laird method.

Table 2
Characteristics of studies.

Author, Year of publication	Study design	Diagnoses methods	Study period	Area	Continents
(Meikle et al., 1999)(20)	Retrospective case studies	enzymatic assay	January 1 1980-December 31 1996	Australia	Oceania
(Poorthuis et al., 1999)(30)	Retrospective study	enzymatic assay	1970–1996	Netherlands	Europe
(Applegarth et al., 2000)(21)	Unclear	enzymatic assay/molecular analysis	1972–1996	British Columbia	North America
(Dionisi-Vici et al., 2002) (22)	Retrospective study	enzymatic assay/molecular analysis	January 1 1985-December 31 1997	Italy	Europe
(Asuman Ozkara and Topcu, 2004) (23)	Records from a list of sources	enzymatic assay	1997–2002	Turkey	Europe
(Revest et al., 2009)(34)	Retrospective study	enzymatic assay/molecular analysis	Unclear	France	Europe
(Poupetova et al., 2010)(31)	Retrospective study	enzymatic assay/molecular analysis	1975-2008	Czech Republic	Europe
(Giraldo et al., 2012)(35)	Retrospective study	enzymatic assay	1976-2002	Iberian Peninsula	Europe
(Mechtler et al., 2012)(24)	Prospective nationwide screening	enzymatic assay/molecular analysis	January 2010-July 2010	Austria	Europe
(Stirnemann et al., 2012)(25)	Retrospective study	enzymatic assay	1980-2010	France	Europe
(Liao et al., 2014)(36)	Unclear	enzymatic assay/molecular analysis	September 2011-January 2013	Taiwan	Asia
(Hult et al., 2014)(26)	Retrospective study	enzymatic assay	1990-2009	Sweden	Europe
(Hopkins et al., 2015)(27)	Pilot study	enzymatic assay/molecular analysis	January 11, 2013-June 11,2013	Missouri	North America
(Burton et al., 2017)(37)	Unclear	enzymatic assay/molecular analysis	November 1 2014-August 31 2016	Illinois	North America

Author, Year of publication	Study design	Diagnoses methods	Study period	Area	Continents
(Kang et al., 2017)(38)	Unclear	enzymatic assay/molecular analysis	Unclear	China	Asia
(Hopkins et al., 2018)(39)	Prospective study	enzymatic assay/molecular analysis	January 11 2013-January 10 2017	Missouri	North America
(Burlina et al., 2018)(40)	Retrospective study	enzymatic assay/molecular analysis	September 2015-January 2017	North East Italy	Europe
(Chien et al., 2020)(28)	Retrospective study	enzymatic assay/molecular analysis	March 2018-April 2019	Taiwan	Asia

Birth prevalence data of GD were extracted from 16 studies and covered 14 areas/countries. The global birth prevalence of GD was 1.5 cases (95% CI: 1.0-2.0) per 100,000 live births. Birth prevalence of GD in Oceania, Europe, North America and Asia were 1.8 cases (95%CI:1.4-2.1), 1.7 cases (95□CI:1.0-2.3), 1.3 cases (95□CI:0.2-2.4) and 1.1 cases (95□CI:-0.1-2.3) per 100,000 live births, respectively (Figure 2).

Two studies that were included for birth prevalence of subtypes of GD were all from Europe (France and Italy). The birth prevalence of GD type 1 (GD 1), type 2 (GD 2) and type 3 (GD 3) were 1.5 cases (95% CI: 1.4-1.7), 0.2 (95% CI: 0.1-0.2) and 0.1 (95% CI: 0.1-0.1) per 100,000 live births, respectively (Figure 3).

Prevalence data of GD were extracted from 4 studies and covered 3 areas/countries. The global prevalence of GD was 0.9 cases (95% CI: 0.7-1.1) per 100,000 inhabitants. Prevalence of GD in Oceania and Europe were 1.7 cases (95%CI: 1.3-2.0) and 0.7 cases (95□CI: 0.7-0.8) per 100,000 inhabitants, respectively (Figure 4).

Although the range of birth prevalence and prevalence of GD was large, no qualitative difference in study methodology that could justify its impact on the pooled estimates was observed. No publication bias was found based on funnel plot and Begg's test for birth prevalence and prevalence of GD (*P* value=0.274 and 0.389) (Figure 5).

Discussion

The upper limit in defining of rare disease ranges from 5 to 76 cases per 100,000 people (10). According to the definition of rare disease, quantitative data by evaluating global epidemiology of GD in this study (1.5 cases (95% CI: 1.0-2.0) per 100,000 live births) confirmed that GD was a rare disease (11). GD is extremely common in Ashkenazi Jews. Goldblatt and Beighton (1979) (12) reported that prevalence of GD in South African Jewish population would be 1:4000. Though population-based genetic screening programs, birth prevalence of GD was predicted to be 1:450 (13). In Ashkenazi Jews, GD may not be considered as rare disease; however, epidemiology data of GD could not represent other races.

Due to founder effect, the data from a specific population from one area/country may affect accuracy of global epidemiology data of GD, so three studies that focused on one specific population were excluded: 1) Goldblatt and Beighton (1979) (12) reported prevalence of GD in Jewish population (1 case in 4000); 2) Swart et al. (1987) (14) reported prevalence of GD in Cape Coloured population (1 case in 247350). 3) Miyamoto et al. (2021) (15) screened 3 GD patients in 5257 people (~90% reported as African-American).

When studies were screening and quality assessing, “incidence” was misused to present frequency of GD among births. It is easy to distinguish the difference between incidence and prevalence. The numerator and denominator of incidence are the number of disease onsets and number of healthy individuals (a population at risk) during periods of observation. The numerator and denominator of prevalence are total number of cases and number of population at a certain moment (16). For new born screening of genetic diseases, including GD, patients were already there, so incidence is not suitable for frequency of GD among births. Birth prevalence, the prevalence at birth, was more suitable to present frequency of GD among births (8, 17).

Theoretically, prevalence should be not far from birth prevalence (18). In this review, birth prevalence of GD (1.5 cases (95% CI: 1.0-2.0) per 100,000 live births) was higher than prevalence of GD (0.9 cases (95% CI: 0.7-1.1) per 100,000 inhabitants). Following reasons could explain such phenomenon: 1) it is very hard to find all GD patients in a population; 2) life span of GD patients is not long enough as normal person.(19) Although birth prevalence was affected by many factors, including diagnostic technology, prenatal diagnosis and termination of pregnancy, birth prevalence of GD may be more accurate than prevalence to calculate the number of GD patients.

To our best knowledge, there was only one comprehensive review of the literature to represent GD epidemiology (5). In this review, author used incidence and birth prevalence at the same time to show frequency of GD among births, which would make reader confused. In the part of “incidence”, 11 studies were used to review “incidence” of GD (5). Among these 11 studies, 8 studies were included in this review to calculate birth prevalence of GD (20–27). The latest study in part of “incidence” that was included in review of Nalysnyk et al. (2017) (27) was published in 2015. The latest study in this review was published in 2020 (28). In the part of prevalence, prevalence of GD was reviewed based on 9 studies (5). Among these 9 studies, 3 and 2 studies were included in this review to calculate prevalence (20, 25, 29) and birth prevalence of GD (30, 31), respectively.

Pooled birth prevalence of GD in Europe was lower than in Oceania; however, the highest birth prevalence of GD was reported in Austria from Europe (5.8 cases per 100,000 live births) (24). The lowest birth prevalence of GD in Europe, 0.2 cases per 100,000 live births, was reported in Turkey (23). The big difference of birth prevalence of GD between Austria and Turkey could be explained by proportion of Ashkenazi Jews (12, 13). Three studies of Asia were all from China, which has low proportion of Ashkenazi Jews. If pooled birth prevalence of GD in Asia contained data from West Asia, the birth prevalence of GD in Asia may be higher.

According to pooled birth prevalence of three subtypes of GD, proportion of patients with GD 1 is about 83% in total patients with GD, which is consistent with review of Stirnemann et al. (2017) (prevalence of GD1: 90–95% in Europe and North America) (1). There were two other studies reported birth prevalence data of three subtypes of GD; however, cases of GD 1 patients were separated to two groups (early and late), meanwhile

cases of GD 2 patients and GD 3 patients were mixed (30, 31). These two studies were excluded to calculate birth prevalence of subtypes of GD.

Quantitative data of global epidemiology of GD could be the fundamental to evaluate the global efforts that improve many factors, including diagnostic technology and data collection, which affect global epidemiology of GD. Life expectancy would be an example to clarify this point. Life expectancy has increased by more than 6 years between 2000 and 2019-from 66.8 years in 2000 to 73.4 years in 2019, globally (32). Life expectancy (from 66.8 years in 2000 to 73.4 years in 2019) of whole-world is pooled global data to show the global efforts to expand life expectancy of citizens worldwide. Global efforts, including development of medical technology, food supply and reduction of war, were carried out not only by one government but also by many governments working together. Unfortunately, life expectancy varies broadly in different countries in 2019, from 50.75 years in Lesotho to 84.26 years in Japan (33). The broadly varied data of life expectancy did not reduce effect that higher pooled global data could reflect global efforts to expand life expectancy of citizens worldwide.

Limitations

There were several limitations of this report: 1) less than 30% studies was assessed as high-quality, highlighting the need for high-quality study about epidemiological evidence of GD; 2) more than half of studies were from Europe (56%). Reports from other continents were underrepresented, which might cause bias to calculate global epidemiology of GD.

Conclusions

To our best knowledge, this is the first systematic review to present quantitative data of global epidemiology of GD. Quantitative data of global epidemiology of GD could be the fundamental to evaluate the global efforts that improve many factors, including diagnostic technology and data collection, which affect global epidemiology of GD.

Abbreviations

GD: Gaucher disease

PRISMA: Preferred Reporting Items for Systematic Reviews and Meta-Analyses

STROBE: STrengthening the Reporting of OBservational studies in Epidemiology

Declarations

Ethics approval and consent to participate

Not applicable.

Consent for publication

Not applicable.

Availability of data and materials

Data from patients can be made available from the corresponding author after discussion with the Institutional Review Board.

Competing interests

The authors declare that they have no competing interests.

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Authors' contributions

MM W searched and collected related articles, then wrote the manuscript. J Z collected related articles. MM W and FQ L separately reviewed all related articles and operated quality assessment. C L and Y M conducted the data analysis. Collection and screening of articles, data analysis and revised writing of the article were supervised by DYX and WJ K. All authors read and approved the final manuscript.

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Figures

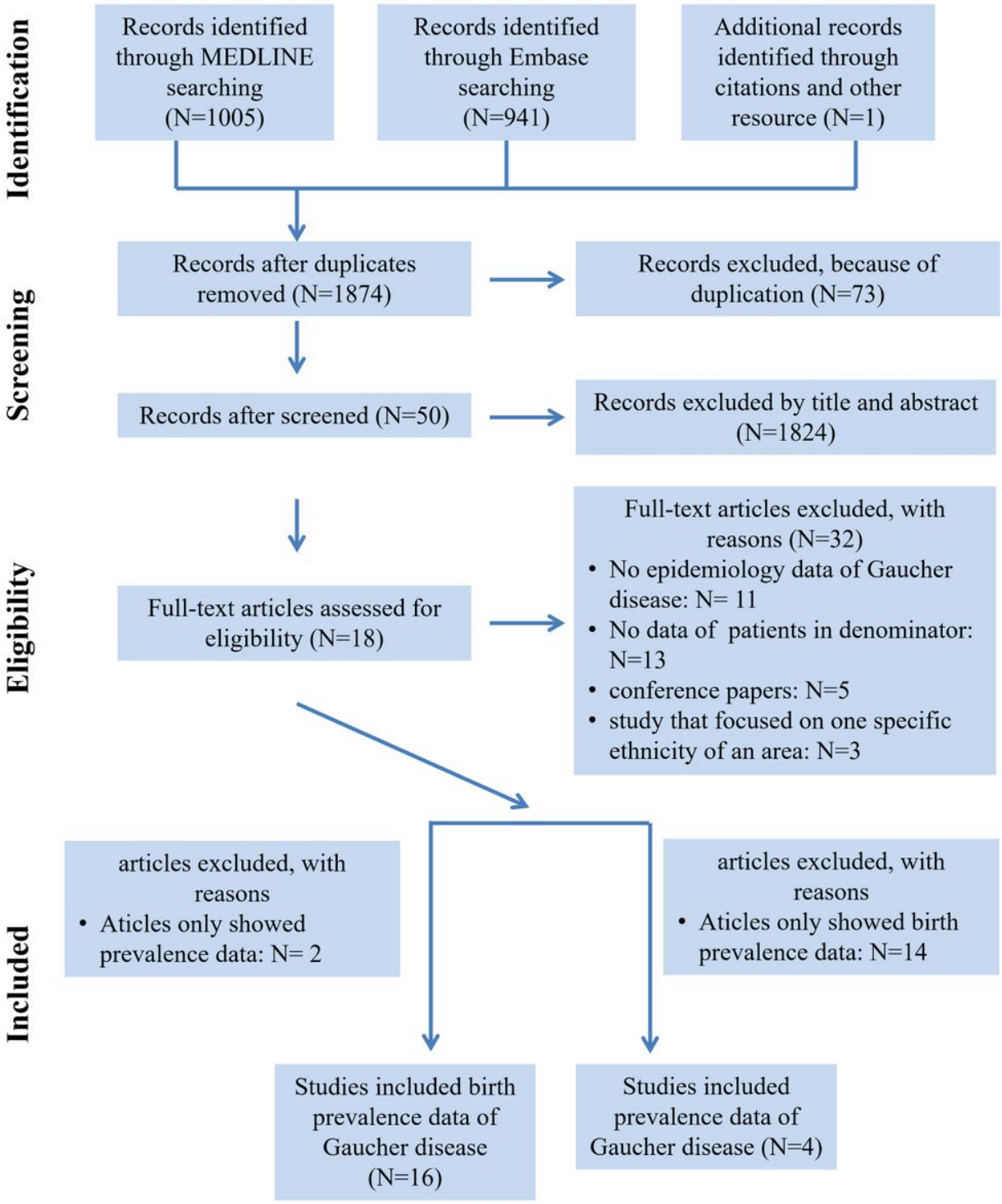

Figure 1

PRISMA flow-chart showing the process of literature search and study selection.

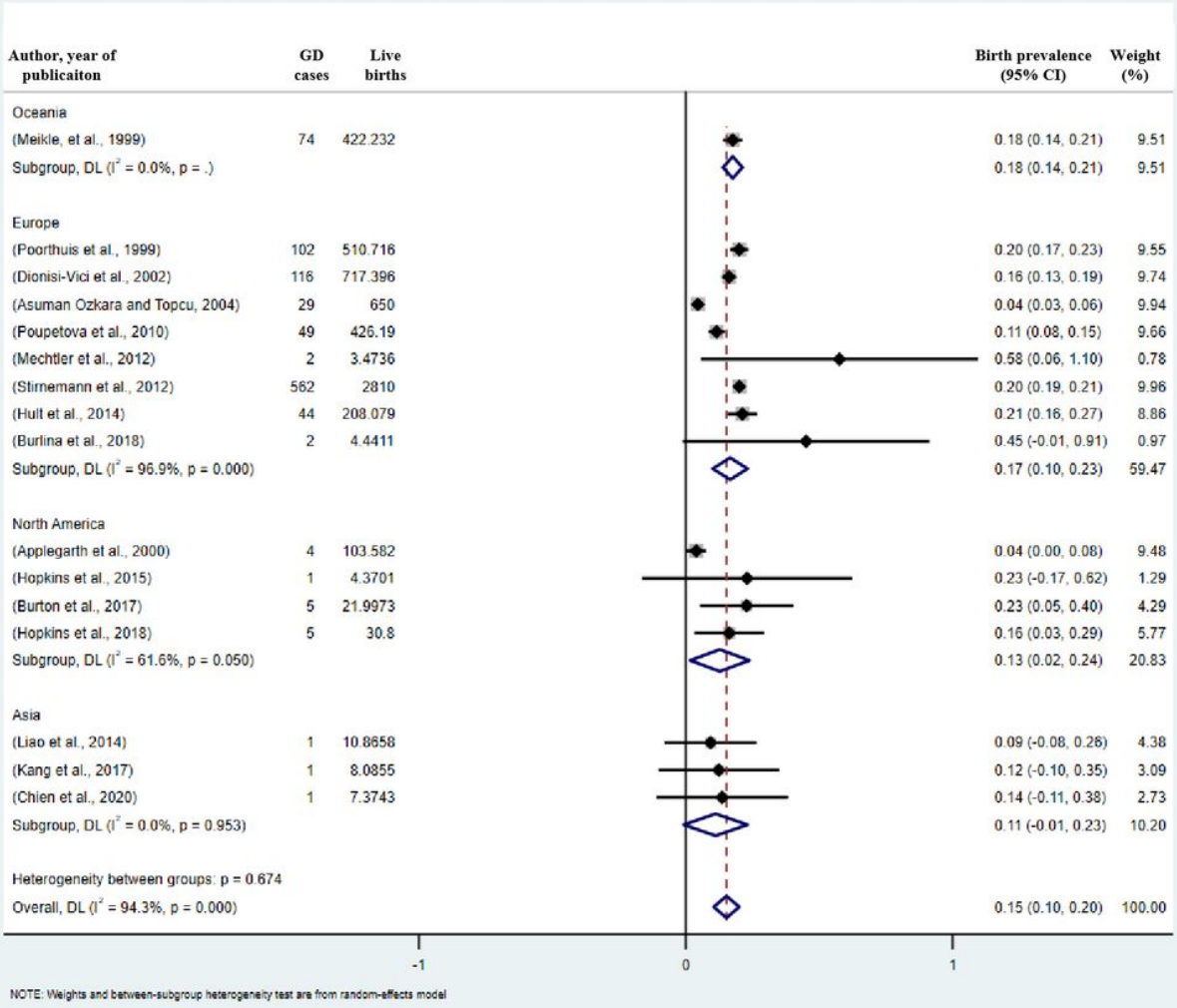

Figure 2

Forest plot of the estimated birth prevalence of Gaucher disease (GD) per 100,000 cases along with 95% confidence interval. NOTE: Weights and between-subgroup heterogeneity test are from random-effects model.

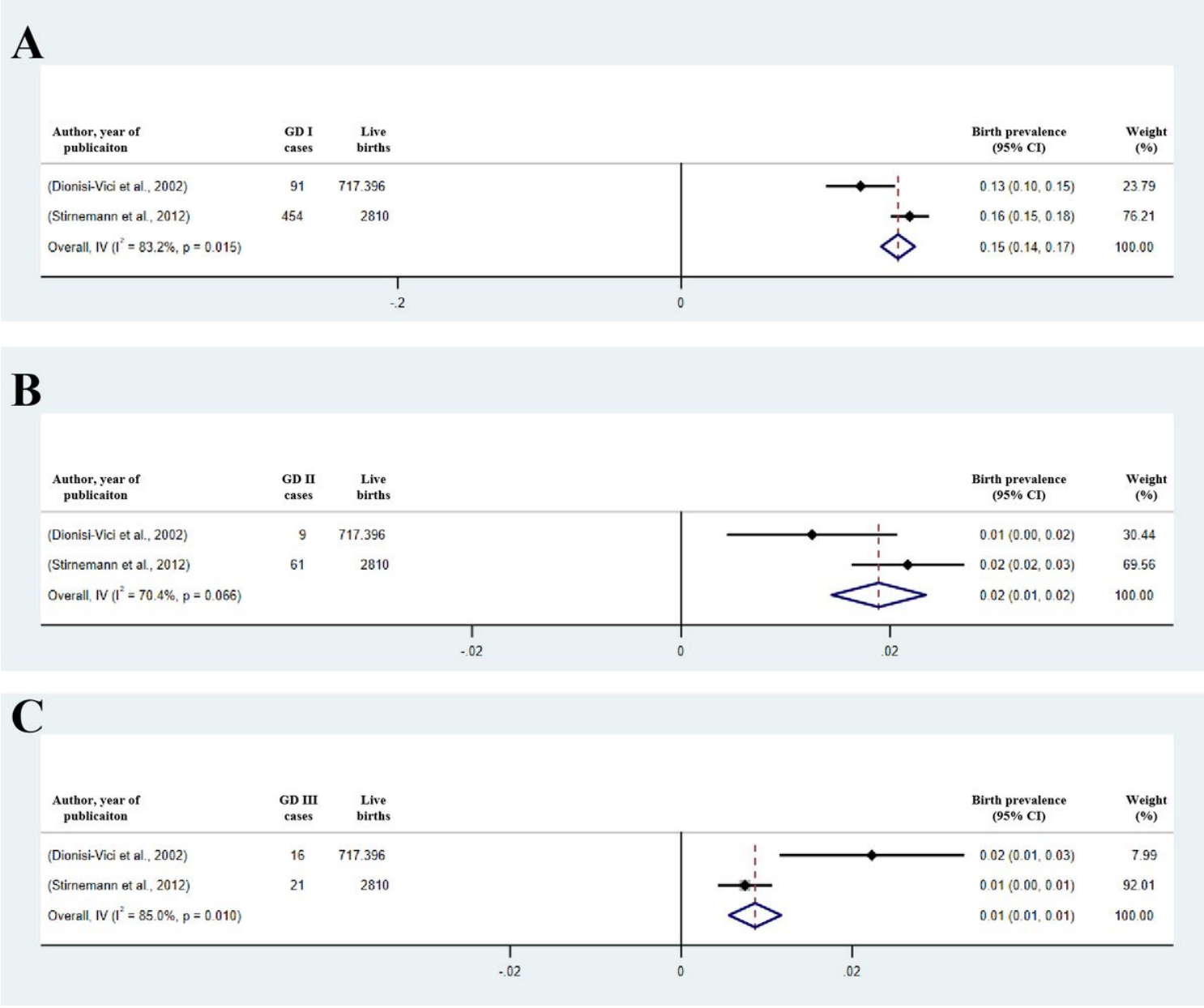

Figure 3

Forest plot of the estimated prevalence of Gaucher disease (GD) per 100,000 cases along with 95% confidence interval. NOTE: Weights and between-subgroup heterogeneity test are from random-effects model.

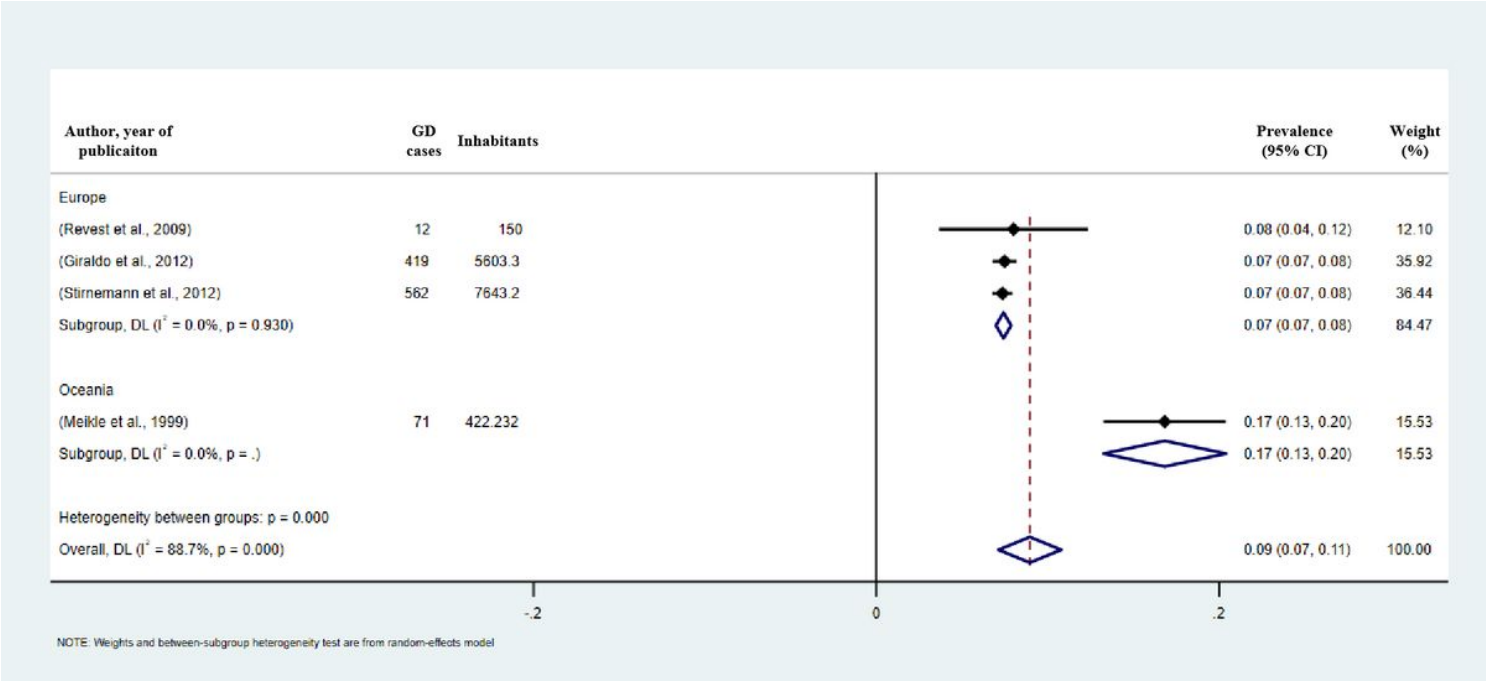

Figure 4

Forest plot of the estimated birth prevalence of Gaucher disease (GD) type 1 (A), type 2 (B) and type 3 (C) per 100,000 cases along with 95% confidence interval. NOTE: Weights and between-subgroup heterogeneity test are from random-effects model.

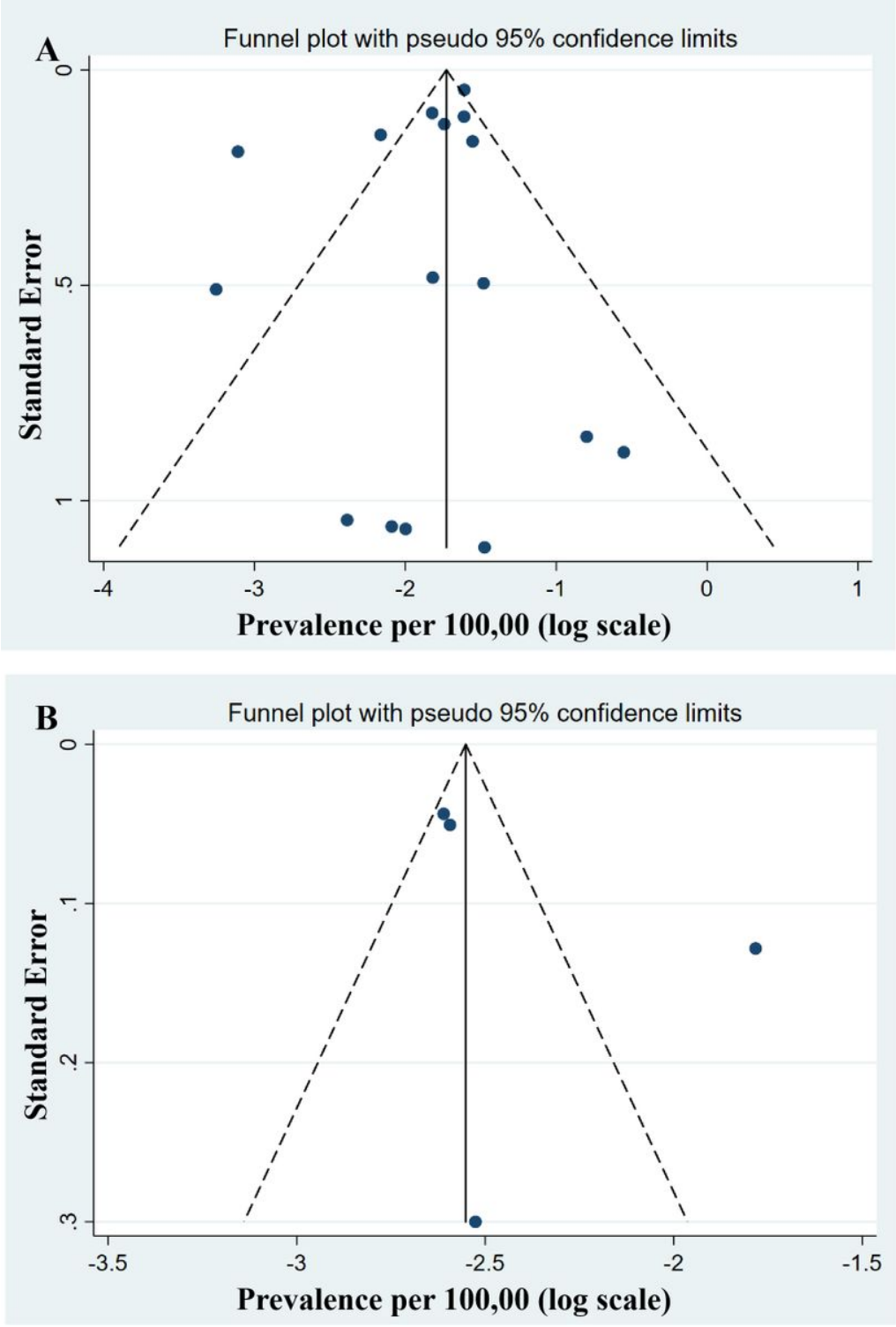

Figure 5

Funnel plot for the estimated birth prevalence (A) and prevalence (B) of Gaucher disease.

Supplementary Files

This is a list of supplementary files associated with this preprint. Click to download.

- Additionalfile1.PRISMAchecklist.doc
- Additionalfile2Literaturesearchstrategy.docx
- Additionalfile3Detailsofqualityassessment.xlsx