INPLASY

Molecular diagnostic yield of next generation sequencing and high resolution melting analysisin osteogenesis imperfecta: a systematic review and meta-analysis

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ADMINISTRATIVE INFORMATION

Support - None.

Review Stage at time of this submission - The review has not yet started.

Conflicts of interest - None declared.

INPLASY registration number: INPLASY202410009

Amendments - This protocol was registered with the International Platform of Registered Systematic Review and Meta-Analysis Protocols (INPLASY) on 04 January 2024 and was last updated on 04 January 2024.

INTRODUCTION

Review question / Objective To investigate the diagnostic yield of next-generation sequencing and high resolution melting analysis in osteogenesis imperfecta.

Condition being studied Osteogenesis imperfecta (OI) is a genetic disorder of connective tissue affecting bone formation and strength, resulting in susceptibility to spontaneous fracture. Diagnosis is based on clinical presentation, family history, radiographic features and genetic analysis. We aim to conduct a systematic review in order to address the question of what percentage of individuals with OI have a positive molecular diagnosis through a next-generation sequencing approach and high resolution melting analysis.

METHODS

Participant or population Articles that met all the following criteria will be included in the meta-analysis: 1) primary research studies, cohort studies, and case series 2) the article uses next generation sequencing or high resolution melting analysis as diagnostic tools for genetic evaluation of patients 3) patients with osteogenesis imperfecta; Articles that met any of the following exclusion criteria will be excluded from the meta-analysis:1) case reports 2) a non-English language article 3) review articles.

Intervention Next generation sequencing.

Comparator Not applicable.

Study designs to be included Articles that met all the following criteria will be included in the meta-analysis: 1) primary research studies, cohort studies, and case series 2) the article uses next generation sequencing or high resolution melting analysis as diagnostic tools for genetic evaluation of patients 3) patients with osteogenesis imperfecta; Articles that met any of the following exclusion criteria will be excluded from the meta-analysis:1) case reports 2) a non-English language article 3) review articles.

Eligibility criteria Not applicable.

Information sources The search will be made in the following databases: PubMed, MEDLINE, EMBASE, Web of Science, and Scopus. Non-English articles will be excluded.

Main outcome(s) The primary outcome is molecular diagnostic yield, which is defined as the number of patients who have variants detected by next-generation sequencing and high resolution melting analysis that are judged to be causative divided by the total number of patients in the cohort.

Additional outcome(s) Not applicable.

Data management Two members on the review team will independently evaluate all potential articles we identify according to our search strategy. Any disagreement will be resolved through discussion or, if necessary, will be decided by a third senior member. Data including the number of patients who had variants detected, the number of total patients included, diagnosis tool, and any other information that needs to be noticed will be manually extracted and recorded into an individual collection for each study, then transferred to Microsoft Excel sheet. Preferred Reporting Items for Systematic Reviews and Meta-analyses (PRISMA) reporting guidelines will be used for assessing data quality.

Quality assessment / Risk of bias analysis Two members on the review team will independently assess the risk of bias. This meta-analysis will follow the Preferred Reporting Items for Systematic Reviews and Meta-analyses (PRISMA) reporting guidelines. This meta-analysis will be evaluated in line with the following criteria: 1) incomplete outcome data (attention bias); 2) selective reporting (reporting bias). The risk of bias will be assessed at three levels for theincluded studies: 1) low risk of bias; 2) unclear risk of bias; 3) high risk of bias.

Strategy of data synthesis Heterogeneity will be quantified by applying the I^2 test with values. The fixed-effects model will be used to incorporate between studies heterogeneity for comparisons with I^2 50%; otherwise, we will conduct use the random-effects logistic regression model.

Subgroup analysis Not applicable.

Sensitivity analysis Heterogeneity will be quantified by applying the I^2 test with values. The fixed-effects model will be used to incorporate between studies heterogeneity for comparisons with I^2 50%; otherwise, we will conduct use the random-effects logistic regression model.

Language restriction Non-English articles will be excluded.

Country(ies) involved China.

Keywords Osteogenesis imperfecta; High-Throughput Nucleotide Sequencing; Humans; Pathology, Molecular.

Contributions of each author

Author 1 - Xuemeng Mu - Acquisition, analysis, interpretation of data, and drafting of the manuscript.
Email: mxm1127@163.com
Author 2 - Hengyan Zhang - Statistical analysis..
Author 3 - Mei Li - Critical review of the manuscript for important intellectual content.
Author 4 - Jia Zhang - Concept and design.