International Platform of Registered Systematic Review and Meta-analysis Protocols

INPLASY

INPLASY202480116

doi: 10.37766/inplasy2024.8.0116

Received: 25 August 2024

Published: 25 August 2024

Corresponding author:
Hassan Vahidnezhad

vahidnezhh@chop.edu

Author Affiliation:
University of Pennsylvania
(Children's Hospital of Philadelphia).

A Systematic Review of Reported Fully Penetrant Sequence Variants and Clinical Characteristics in Association with 117 Patients with Single-gene Pyoderma Gangrenosum

Norouzi-Barough, L; Biglari, S; Sherkat, R; Gudjonsson, JE; Hakonarson, H; Vahidnezhad, H.

ADMINISTRATIVE INFORMATION

Support - Leo Foundation and NIH R01 grant.

Review Stage at time of this submission - Piloting of the study selection process.

Conflicts of interest - None declared.

INPLASY registration number: INPLASY202480116

Amendments - This protocol was registered with the International Platform of Registered Systematic Review and Meta-Analysis Protocols (INPLASY) on 25 August 2024 and was last updated on 25 August 2024.

INTRODUCTION

Review question / Objective What are the fully penetrant sequence variants associated with single-gene pyoderma gangrenosum? How do these genetic variants correlate with the clinical characteristics observed in reported cases?

Condition being studied Pyoderma gangrenosum (PG) is a rare cutaneous inflammatory disorder classified within the spectrum of neutrophil-mediated skin diseases known as neutrophilic dermatoses (ND) with a poorly understood etiology. NDs are typically characterized by aberrant and prominent infiltration of sterile neutrophils in different layers of the affected skin, including the epidermis, dermis, or even the hypodermis, without any evidence of infection.

METHODS

Participant or population People whose clinical diagnosis and pathology are Pyoderma gangrenosum (PG).

Intervention Not applicable.

Comparator Not applicable.

Study designs to be included We will include randomised trials, cohort, Case report, Case series, Cross-sectional, letter to the editor andcase– control studies.

Eligibility criteria Our exclusion criteria will study without genetic investigations or those that did not report variant/mutation in the suspected gene, PG-associated autoinflammatory syndromes lacking

Pyoderma gangrenosum (PG) presentations, and non-English language publications.

Information sources We will conduct a systematic search on major electronic bibliographic databases (MEDLINE, Web of Science, Scopus, and Google Scholar).

Main outcome(s) Identifying genes involved in genetic defects associated with Pyoderma gangrenosum (PG) and providing suggested methods for the diagnosis of Pyoderma gangrenosum (PG) RW patients.

Quality assessment / Risk of bias analysis The tool developed by Joanna Briggs Institute (Adelaide, Australia) (http://joannabriggs.org) for quality assessments for case reports, case series, and cross-sectional research will use.

Strategy of data synthesis The following data, if available, will extract from the included references: (i) Strict phenotypic criteria of Pyoderma gangrenosum (PG), (ii) Causative gene, (iii) Inheritance, (iv) Associated features, (v) Immunological features, and (vi) infection type. We will provide a narrative synthesis of the findings from the included studies, structured around the type of Based on the IUIS classification, we will determine the category of each gene to determine the immune pathways involved.

Subgroup analysis Not applicable.

Sensitivity analysis Not applicable.

Country(ies) involved Iran.

Keywords Pyoderma gangrenosum, Neutrophilic dermatosis, Inborn errors of immunity, Mendelian mutations, Monogenic disorder.

Contributions of each author

Author 1 - Leyla Norouzi-Barough.
Email: l.norouzi61@gmail.com
Author 2 - Sajjad Biglari.
Email: s1369b@yahoo.com
Author 3 - Roya Sherkat.
Author 4 - Johann E. Gudjonsson.
Author 5 - Hakon Hakonarson.
Author 6 - Hassan Vahidnezhad.
Email: vahidnezhh@chop.edu