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Monogenic etiologies of epidermodysplasia verruciformis: A comprehensive systematic review

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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.

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Amendments - This protocol was registered with the International Platform of Registered Systematic Review and Meta-Analysis Protocols (INPLASY) on 02 October 2023 and was last updated on 02 October 2023.

INTRODUCTION

Review question / Objective Is epidermodysplasia verruciformis associated with any Mendelian disorders?

Condition being studied Epidermodysplasia verruciformis is a rare autosomal recessive genetic skin disorder characterized by an exclusive susceptibility to cutaneous HPV in the absence of other infections. Approximately 30–40% of Epidermodysplasia verruciformis patients develop non-melanoma skin cancer two to three decades after the onset of Epidermodysplasia verruciformis. Epidermodysplasia verruciformis can be further categorized into typical, atypical, and acquired types.

METHODS

Participant or population Patients with epidermodysplasia verruciformis phenotype.

Intervention None.

Comparator None.

Study designs to be included Letters to the editor, case series, case reports, cross-sectional studies, clinical trials, and case-control studies.

Eligibility criteria Articles will include when the diagnosis of EV was linked to a single-gene etiology. Articles will exclude from our study if they didn't mention any specific causal gene; articles with non-monogenic causality for EV, including

multigene deletions, duplications, and aneuploidies; articles with acquired EV subjects; articles that didn't provide enough information about the patient's phenotype or clinical information.

Information sources MEDLINE, Scopus, Web of Science (science and social science citation index), and Google Scholar. Hand and citation searching.

Main outcome(s) Identifying genes with pathogenic variants that cause epidermodysplasia verruciformis.

Quality assessment / Risk of bias analysis Quality assessment tools for case reports, case series, and cross-sectional studies are available from the Joanna Briggs Institute (Adelaide, Australia) (http://joannabriggs.org/). Each study will be classified into one of the following groups: (i) good if all quality criteria were judged as “present,” (ii) fair if one or more key domains were “unclear,” or (iii) poor if one or more key domains were “absent”.

Strategy of data synthesis The following data will be retrieved from the included papers: (i) phenotypes of Epidermodysplasia verruciformis and other co-morbidities; (ii) causal gene; (iii) inheritance; (iv) number of patients; (v) immunological features; (vi) age-of-onset; (vii) cancers.

Subgroup analysis This is a qualitative synthesis and while subgroup analyses may be undertaken it is not possible to specify the groups in advance.

Sensitivity analysis This is a qualitative synthesis and it is not applicable.

Language restriction English.

Country(ies) involved United States.

Keywords epidermodysplasia verruciformis, HPV, inborn errors of immunity, persistent human papillomavirus infection, monogenic disorderepidermodysplasia verruciformis, HPV, inborn errors of immunity.

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