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Clinical characteristics, therapeutic options and outcomes in Camurati-Engelmann disease: a systematic review

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ADMINISTRATIVE INFORMATION

Support - None.

Review Stage at time of this submission - Preliminary searches.

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 29 December 2023 and was last updated on 29 December 2023.

INTRODUCTION

Review question / Objective To describe the varied manifestations of Camurati-Engelmann disease; Describe the various treatment agents used; Therapeutic response to these agents.

Rationale Given the rarity of the disease with only about 400 cases reported in literature, the experience in managing this condition is limited. While corticosteroids do help in relieving pain, there are concerns about worsening osteoporosis with its long-term use. Losartan which is an angiotensin receptor antagonist inhibits TGFβ1 signalling and has been used in CED with variable efficacy. Other agents that have been used in literature include bisphosphonates, non-steroidal anti-inflammatory agents, acetaminophen and calcitonin. This systematic review is being undertaken to comprehensively describe the varied manifestations, clinical characteristics, treatment modalities used and therapeutic response to these agents as reported in literature till date.

Condition being studied Camurati-Engelmann disease (CED) also known as progressive diaphyseal dysplasia is characterized by hyperostosis of the long bones and skull and other manifestations such as proximal muscle weakness, a waddling gait. Joint contractures, macrocephaly and occasional cranial nerve impingement. A rare disease, it is inherited in an autosomal dominant fashion and is due to activating mutations of the transforming growth factor beta (TGFβ1) gene. The diaphysis of long tubular bones and the skull are usually involved; the metaphyseal region and the spine are usually spared. Activation of TGFβ1 results in increased differentiation of osteoblasts as compared to osteoclasts. Intramembranous

ossification which is regulated by the activities of osteoblasts and osteoclasts plays a role in the ossification of the skull and diaphysis of long bones; this explains the characteristic sites of involvement in CED.

METHODS

Search strategy A systematic search of the literature published in English language will be performed in keeping with the Preferred Reporting Items for Systematic Review and Meta-Analysis (PRISMA) statement. Pubmed, and Scopus, databases will be searched from articles published through December 2023-January 31, 2024 evaluating the outcome of various treatment agents in Camurati-Engelmann disease.

Participant or population Patients diagnosed with Camurati-Engelmann disease.

Intervention Various treatment agents.

Comparator None.

Study designs to be included Case reports and case series.

Eligibility criteria All papers published in the English language with a diagnosis of Camurati-Engelmann disease and treatment outcomes following various agents will be included. The exclusion criteria comprised studies primarily written in a language other than English, narrative reviews, conference abstracts and patients having other sclerotic bone diseases.

Information sources Pubmed, Scopus.

Main outcome(s) Symptomatic improvement of patients; Whether any progression of disease or disease remains stable.

Quality assessment / Risk of bias analysis All full texts were read to evaluate the methodological quality of the included cases reports. Then the authors will use a recently proposed tool, based on the previous criteria from the Pierson, Bradford Hills, and Newcastle Ottawa Scale, and categorized into 4 domains: selection, ascertainment, causality, and reporting. An overall judgement will be made about methodological quality as follows (1: medium; 2: good; 3: very good; 4: excellent). The quality's appraisal will be conducted by one author (KEC) and verified by a second author (TVP). Any discrepancy will be resolved by consensus.

Strategy of data synthesis Data will be summarized using descriptive statistics, with means and standard deviations for continuous variables and frequencies and percentages for dichotomous variables. When the data are non-normally distributed, they will be described as median with inter-quartile ranges as appropriate.

Subgroup analysis Will be decided after tabulation of data.

Sensitivity analysis NA.

Language restriction English language only.

Country(ies) involved India.

Keywords Camurati-Engelmann disease, Progressive diaphyseal dysplasia, corticosteroids, losartan, bisphosphonates.

Contributions of each author

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