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INPLASY

# RRM2B-Associated Chronic Progressive External Ophthalmoplegia: A Case Report and Systematic Review of Literature

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## ADMINISTRATIVE INFORMATION

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**Review Stage at time of this submission -** Completed but not published.

**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 12 August 2024 and was last updated on 12 August 2024.

## INTRODUCTION

**Review question / Objective** What are the general characteristics of RRM2B mutation-associated mitochondrial diseases? What are common presenting symptoms, inheritance patterns, diagnosis and genetic testing results of patients diagnosed with RRM2B mutations-associated mitochondrial disease?
Population : patients with RRM2B mutations
Intervention : N/A
Comparison : other patients with RRM2B mutations
Outcome : Diagnosis, genetic testing results

**Rationale** To our understanding, a systematic review on RRM2B mutations does not currently exist. By compiling and analyzing the current cases published describing patients presenting with RRM2B mutations, our review will provide a comprehensive resource for physicians and scientists encountering RRM2B-associated mitochondrial diseases.

**Condition being studied** RRM2B-Associated Chronic Progressive External Ophthalmoplegia (RRM2B-CPEO) is a mitochondrial disease that results from mutations to a protein responsible for DNA repair. Few cases of RRM2B-CPEO have been reported, and the clinical spectrum (which includes multiple organ systems) has not been comprehensively described.

## METHODS

**Search strategy** The review followed the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) guidelines. An extensive search of PubMed was done using the search terms:
“RRM2B” and “mitochondrial disease”
“RRM2B” and “Kearns-Sayre”
“RRM2B” and“CPEO”.

**Participant or population** Patients with RRM2B mutation-associated disease.

**Intervention** Not applicable.

**Comparator** Not applicable.

**Study designs to be included** All types of studies will be included if they describe individual cases of patients with RRM2B mutations.

**Eligibility criteria** Inclusion criteria: patients with RRM2B mutation.
Exclusion criteria: lack of cases present and/or no RRM2B mutations.

**Information sources** An electronic database (Pubmed) will be used as an information source.

**Main outcome(s)** Main outcomes will include age at presentation, sex, presenting symptoms, inheritance pattern, diagnosis, and genetic testing results. Each variable will be extracted at the time of data collection.

**Additional outcome(s)** Not applicable.

**Quality assessment / Risk of bias analysis** No formal tools will be utilized, but risk of bias will be discussed in the Discussion portion of the manuscript.

**Strategy of data synthesis** When applicable, data will be synthesized using percentages, means, and standard deviations.

**Subgroup analysis** There are no planned subgroup investigations.

**Sensitivity analysis** One reviewer will independently perform the initial search with key terms. The same reviewer will read the title and abstract of each search result to screen for inclusion criteria. If there is uncertainty whether the paper meets inclusion criteria based on the title and abstract, the reviewer will access the full paper to determine whether criteria is met. If there is still uncertainty, the reviewer will consult a second reviewer to assist with the decision.

**Language restriction** Yes: only papers with an accessible english version will be included.

**Country(ies) involved** United States of America.

**Keywords** RRM2B ; chronic progressive external ophthalmoplegia ; CPEO ; mitochondrial disorders.

**Dissemination plans** This systematic review will be submitted to a peer-reviewed journal for publication.

**Contributions of each author**

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