Exome and genome sequencing for pediatric patients with congenital anomalies or intellectual disability: an evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG).

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American College of Medical Genetics and Genomics (ACMG)

Manickam K, McClain MR, Demmer LA, et al. Exome and genome sequencing for pediatric patients with congenital anomalies or intellectual disability: an evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2021 Nov;23(11):2029-2037. [93 references] PubMed

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Overview

Guideline Objective

To develop an evidence-based clinical practice guideline for the use of exome and genome sequencing (ES/GS) in the care of pediatric patients with one or more congenital anomalies (CA) or intellectual disability (ID)

Patient Population

Pediatric patients with one or more CA with onset prior to age 1 year or developmental delay (DD) or ID with onset prior to age 18 years

Recommendations

Recommendation Statements

Major interventions covered in this guideline include:

- Use of ES and GS as a first-tier or second-tier test after chromosomal microarray (CMA) or focused testing

Note: Full recommendation statements have not been provided because this guideline does not meet EGT's systematic review of the evidence criteria. Refer to the original guideline for more information.

Evidence Rating Scheme

Refer to the original guideline documentation for more information.

Recommendation Rating Scheme

Refer to the original guideline documentation for more information.

Related Content

Supporting Documents

- Systematic Evidence-based Review: Outcomes from Exome and Genome Sequencing for Pediatric Patients with Congenital Anomalies or Intellectual Disability; 2020 Jun.

Implementation Tools

No implementation tools available.

Patient Education

No patient education materials available.

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