The topic, Genetic Testing for Developmental Diseases, is not feasible for a full systematic review due to the limited data available at this time; however, it will be considered for a potential technical brief by the Effective Health Care (EHC) Program.

To see a description of a technical brief, please go to http://effectivehealthcare.ahrq.gov/index.cfm/research-for-policymakers-researchers-and-others/.

If this topic is developed into a technical brief, a protocol will be posted on the AHRQ Web site. To sign up for notification when this and other EHC Program topics are posted, please go to http://effectivehealthcare.ahrq.gov/index.cfm/join-the-email-list1/.

### Topic Description

**Nominator(s):** Public payer

**Nomination Summary:**

The nomination is concerned about increased ordering of advanced genetic testing (comparative genomic hybridization [CGH], chromosomal microarray analysis [CMA] and array CGH [aCGH]) to identify causes of developmental and intellectual disabilities (DD/ID) or autism spectrum disorder (ASD) in children and is interested to know the clinical utility of these sophisticated genetic tests.

**Staff-Generated PICO**

- **Population(s):** Children with developmental or intellectual (DD/ID) disabilities and autism spectrum disorder (ASD)
- **Intervention(s):** Chromosomal microarray analysis/other genetic testing
- **Comparator(s):** Other genetic testing/no genetic testing
- **Outcome(s):** Changes to child treatment specific to genetic etiology that results in improved outcomes

**Key Questions from Nominator:**

- Does the identification of the genetic etiology for DD/ID or ASD improve treatment?
- Which children should be tested?
- Which genetic tests should be conducted?
Considerations

- The topic meets EHC Program appropriateness and importance criteria. (For more information, see [http://effectivehealthcare.ahrq.gov/index.cfm/submit-a-suggestion-for-research/how-are-research-topics-chosen/](http://effectivehealthcare.ahrq.gov/index.cfm/submit-a-suggestion-for-research/how-are-research-topics-chosen/).)

- Children with neurodevelopmental delays during their early years may eventually be diagnosed with DD/ID and/or ASD. These lifelong conditions may place limits on affected children's functional abilities and adaptive behavior. Their potential for independent living may also be limited based upon their specific condition and severity.

- Diagnosis of children with DD/ID or ASD can often be made based upon the clinical history and physical examination. Identifying genetic abnormalities is more complicated in children who are without syndromic features, too young for full expression of condition, or with atypical presentations. Identifying a specific genetic abnormality in these cases may provide an explanation for a child's DD/ID.

- There is currently not enough evidence that links genetic testing to meaningful treatment changes in decisions and patient centered outcomes. Most of the evidence looks at diagnostic yield, not clinical outcomes.

- Given the limited available evidence, the rapid pace of change in genetic testing, and the recent availability of even more sophisticated genetic testing for DD/ID and ASD, a technical brief that examines available tests may provide a useful summary and could highlight where there are gaps in the research.