# Genetic testing for global developmental delay

## Measure Description
Percentage of patients who had genetic testing ordered for global developmental delay (GDD)

## Measure Components

<table>
<thead>
<tr>
<th><strong>Numerator Statement</strong></th>
<th>Patients for whom chromosomal microarray (CMA) was ordered.</th>
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</thead>
<tbody>
<tr>
<td><strong>Denominator Statement</strong></td>
<td>All children less than 6 years of age with GDD* of unknown etiology</td>
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<tr>
<td>*GDD defined as developmental skills of more than 2 Standard Deviations below age-matched peers in 2 or more aspects of the 5 domains of development (motor, speech and language, cognitive, social, adaptive).</td>
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| **Denominator Exceptions** | - Patient/caregiver refuse  
- Referred to or under the care of a geneticist |
| **Exception Justification** | Parental/caregiver approval is necessary in proceeding with genetic testing. If a patient has an established geneticist providing services, additional evaluation is redundant and burdensome. |
| **Supporting Guideline & Other References** | The following statements are quoted verbatim from the referenced supporting articles:  
- “Microarray testing is abnormal on average in 7.8% of subjects with GDD/ID and in 10.6% of those with syndromic features (Class III).”1  
- “Karyotype studies are abnormal in at least 4% of subjects with GDD/ID and in 18.6% of those with syndromic features (Class II and III).”1  
- “StFISH testing is abnormal in at least 3.5% of subjects with GDD/ID, in at least 4.2% of those with syndromic features, in as few as 0.5% of those with mild impairment, and in at least 7.4% of those with moderate/severe impairment (Class I, II, and III).”2  
- “Mutation in X-linked genes may explain up to 10% of all cases of GDD/ID. Testing of XLID genes has a yield of 42% in males from definitely X-linked families and of 17% in males from possibly X-linked families (Class III).”2  
- “MeCP2 mutations are found in 1.5% of girls with moderate/severe GDD/ID and in less than 0.5% of males with GDD/ID (Class III).”1  
- “Screening for IEMs in children with GDD/ID has a yield of between 0.2% and 4.6%, depending on the presence of clinical indicators and the range of testing performed (Class III).”2  
- “Testing for CDGs has a yield of up to 1.4%, and testing for creatine synthesis and transport disorders has a yield of up to 2.8% (Class III).”1  
- “…Confirm the clinical diagnosis with the appropriate genetic testing, as warranted by clinical circumstances.”3  
- “If a specific diagnosis is suspected, arrange for the appropriate diagnostic studies to confirm including single-gene tests or chromosomal microarray test.”3 |
• “If diagnosis is unknown and no clinical diagnosis is strongly suspected, begin the stepwise evaluation process:
  o Chromosomal microarray should be performed in all
  o Fragile X genetic testing should be performed in all”
• “If no diagnosis established:
  o Male gender and family history suggestive X-linkage, complete XLID panel that contains genes causal of nonsyndromic XLID and complete high-density X-CMA. Consider X-inactivation skewing in the mother of the proband.
  o Female gender: complete MECP2 deletion, duplication, and sequencing study”
• “It is important to emphasize the new role of the genomic microarray as a first-line test, as well as the renewal of efforts to identify the child with an inborn error of metabolism.”
• “CMA offers a much higher diagnostic yield (15%-20%) for genetic testing of individuals with unexplained DD/ID, ASD, or MCA than a G-banded karyotype (~3%, excluding Down syndrome and other recognizable chromosomal syndromes), primarily because of its higher sensitivity for submicroscopic deletions and duplications.”
• “Available evidence strongly supports the use of CMA in place of G-banded karyotyping as the first-tier cytogenetic diagnostic test for patients with DD/ID, ASD, or MCA. G-banded karyotype analysis should be reserved for patients with obvious chromosomal syndromes (e.g., Down syndrome), a family history of chromosomal rearrangement, or a history of multiple miscarriages.”

<table>
<thead>
<tr>
<th>Measure Importance</th>
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<tr>
<td><strong>Relationship to Desired Outcome</strong></td>
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<tr>
<td><strong>Opportunity for Improvement</strong></td>
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| **National Quality Strategy Domains** | □ Patient and Family Engagement  
□ Patient Safety  
□ Care Coordination  
□ Population/Public Health  
□ Efficient Use of Healthcare Resources  
☒ Clinical Process/Effectiveness |
<table>
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<tr>
<th>Harmonization with Existing Measures</th>
<th>N/A</th>
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<td><strong>Measure Designation</strong></td>
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| **Measure Purpose** (Check all that apply) | ☒ Quality improvement  
  □ Accountability |
| **Type of Measure** (Check all that apply) | ☒ Process  
  □ Outcome  
  □ Structure |
| **Level of Measurement** (Check all that apply) | ☒ Individual Provider  
  ☒ Practice  
  ☒ System |
| **Care Setting** (Check all that apply) | ☒ Outpatient  
  □ Inpatient  
  □ Emergency Departments and Urgent Care  
  □ Residential (i.e., nursing facility, domiciliary, home care) |
| **Data Source** (Check all that apply) | ☒ Electronic health record (EHR) data  
  □ Administrative Data/Claims  
  ☒ Patient Medical Record  
  ☒ Registry |
| **References**                       |     |
| **Denominator (Eligible Population)** | ICD-10 Code  
  F88.X Global developmental delay  
  AND  
  CPT E/M Service Code  
  **99201, 99202, 99203, 99204, 99205** Office or other outpatient visit 10, 20, 30, 45, or 60 minutes for the evaluation and management of a new patient; |
| 99211, 99212, 99213, 99214, 99215 | Office or other outpatient visit 5, 10, 15, 25, or 40 minutes for the evaluation and management of an established patient |