When Services Are Eligible for Coverage
Coverage for eligible medical treatments or procedures, drugs, devices or biological products may be provided only if:

- Benefits are available in the member’s contract/certificate, and
- Medical necessity criteria and guidelines are met.

Based on review of available data, the Company may consider genetic testing for Rett syndrome (RTT)−associated genes (e.g., methyl-CpG-binding protein 2 [MECP2], forkhead box G1 [FOXG1], or cyclin-dependent kinase-like 5 [CDKL5]) to establish a genetic diagnosis of Rett syndrome (RTT) in a child with developmental delay and signs/symptoms of Rett syndrome (RTT), when a definitive diagnosis cannot be made without genetic testing to be eligible for coverage.**

Based on review of available data, the Company may consider targeted genetic testing for a known familial Rett syndrome (RTT)−associated variant to determine carrier status of a first-degree female relatives (a mother or a sister) of an individual with Rett syndrome (RTT) to be eligible for coverage.**

When Services Are Considered Investigational
Coverage is not available for investigational medical treatments or procedures, drugs, devices or biological products.

Based on review of available data, the Company considers all other indications for genetic testing for Rett syndrome (RTT)−associated genes (eg, MECP2, FOXG1, or CDKL5), including routine carrier testing (preconception or prenatal) in persons with negative family history, and testing of asymptomatic family members to determine future risk of disease, to be investigational.*
Genetic Testing for Rett Syndrome

Policy #: 00369
Original Effective Date: 11/20/2013
Current Effective Date: 01/09/2023

Policy Guidelines
Genetics Nomenclature Update
The Human Genome Variation Society nomenclature is used to report information on variants found in DNA and serves as an international standard in DNA diagnostics. It is being implemented for genetic testing medical evidence review updates starting in 2017 (see Table PG1). The Society's nomenclature is recommended by the Human Variome Project, the HUman Genome Organization, and by the Human Genome Variation Society itself.

The American College of Medical Genetics and Genomics and the Association for Molecular Pathology standards and guidelines for interpretation of sequence variants represent expert opinion from both organizations, in addition to the College of American Pathologists. These recommendations primarily apply to genetic tests used in clinical laboratories, including genotyping, single genes, panels, exomes, and genomes. Table PG2 shows the recommended standard terminology-"pathogenic," "likely pathogenic," "uncertain significance," "likely benign," and "benign"-to describe variants identified that cause Mendelian disorders.

Table PG1. Nomenclature to Report on Variants Found in DNA

<table>
<thead>
<tr>
<th>Previous</th>
<th>Updated</th>
<th>Definition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mutation</td>
<td>Disease-associated</td>
<td>Disease-associated change in the DNA sequence</td>
</tr>
<tr>
<td></td>
<td>variant</td>
<td></td>
</tr>
<tr>
<td>Variant</td>
<td>Change in the DNA sequence</td>
<td></td>
</tr>
<tr>
<td>Familial variant</td>
<td>Disease-associated variant identified in a proband for use in subsequent targeted genetic testing in first-degree relatives</td>
<td></td>
</tr>
</tbody>
</table>

Table PG2. ACMG-AMP Standards and Guidelines for Variant Classification

<table>
<thead>
<tr>
<th>Variant Classification</th>
<th>Definition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pathogenic</td>
<td>Disease-causing change in the DNA sequence</td>
</tr>
<tr>
<td>Likely pathogenic</td>
<td>Likely disease-causing change in the DNA sequence</td>
</tr>
<tr>
<td>Variant of uncertain significance</td>
<td>Change in DNA sequence with uncertain effects on disease</td>
</tr>
</tbody>
</table>
Genetic Testing for Rett Syndrome

Policy # 00369
Original Effective Date: 11/20/2013
Current Effective Date: 01/09/2023

<table>
<thead>
<tr>
<th>Likely benign</th>
<th>Likely benign change in the DNA sequence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Benign</td>
<td>Benign change in the DNA sequence</td>
</tr>
</tbody>
</table>

ACMG: American College of Medical Genetics and Genomics; AMP: Association for Molecular Pathology.

Genetic Counseling
Experts recommend formal genetic counseling for patients who are at risk for inherited disorders and who wish to undergo genetic testing. Interpreting the results of genetic tests and understanding risk factors can be difficult for some patients; genetic counseling helps individuals understand the impact of genetic testing, including the possible effects the test results could have on the individual or their family members. It should be noted that genetic counseling may alter the utilization of genetic testing substantially and may reduce inappropriate testing; further, genetic counseling should be performed by an individual with experience and expertise in genetic medicine and genetic testing methods.

Background/Overview
Rett Syndrome
Rett syndrome (RTT) is a severe neurodevelopmental disorder primarily affecting girls, with an incidence of 1 in 10,000 female births, making it among the most common genetic causes of intellectual disability in girls. In its typical form, RTT is characterized by apparently normal development for the first 6 to 18 months of life, followed by regression of intellectual functioning, acquired fine and gross motor skills, and social skills. Purposeful use of the hands is replaced by repetitive stereotyped hand movements, such as hand-wringing. Other clinical manifestations include seizures, disturbed breathing patterns with hyperventilation and periodic apnea, scoliosis, growth retardation, and gait apraxia.

There is wide variability in the rate of progression and severity of the disease. In addition to the typical (or classic) form of RTT, there are recognized atypical variants. Three distinct atypical variants have been described: preserved speech, early seizure, and congenital variants. RTT occurring in males is also considered a variant type and is associated with somatic mosaicism or Klinefelter (XXY) syndrome. A small number of RTT cases in males arising from the MECP2 exon 1 variant have been reported. Diagnostic criteria for typical (or classic) RTT and atypical (or variant)
RTT have been established. For typical RTT, a period of regression followed by recovery or stabilization and fulfillment of all the main criteria are required to meet the diagnostic criteria for classic RTT. For atypical RTT, a period of regression followed by recovery or stabilization, at least 2 of the 4 main criteria, plus 5 of 11 supportive are required to meet the diagnostic criteria of variant RTT.

**Treatment**
Currently, there are no specific treatments that halt or reverse disease progression, and there are no known medical interventions that will change the outcome of patients with RTT. Management is mainly symptomatic and individualized, focusing on optimizing each individual's abilities. A multidisciplinary approach is usually applied, with specialist input from dietitians, physical therapists, occupational therapists, speech therapists, and music therapists. Regular monitoring for scoliosis (seen in >87% of patients by age 25 years) and possible heart abnormalities, particularly cardiac conduction abnormalities, may be recommended. Spasticity can have a major impact on mobility; physical therapy and hydrotherapy may prolong mobility. Occupational therapy can help children develop communication strategies and skills needed for performing self-directed activities (eg, dressing, feeding, practicing arts and crafts).

Pharmacologic approaches to managing problems associated with RTT include melatonin for sleep disturbances and several agents to control breathing disturbances, seizures, and stereotypic movements. RTT patients have an increased risk of life-threatening arrhythmias associated with a prolonged QT interval, and avoidance of a number of drugs is recommended, including prokinetic agents, antipsychotics, tricyclic antidepressants, antiarrhythmics, anesthetic agents, and certain antibiotics.

In a mouse model of RTT, genetic manipulation of the *MECP2* gene has demonstrated reversibility of the genetic defect.

**Genetics**
RTT is an X-linked dominant genetic disorder. Pathogenic variants in the *MECP2* gene, which is thought to control expression of several genes, including some involved in brain development, were first reported in 1999. Subsequent screening has shown that over 80% of patients with classic RTT have pathogenic variants in the *MECP2* gene. More than 200 pathogenic variants in *MECP2* have been associated with RTT. However, 8 of the most commonly occurring missense and nonsense
variants account for almost 70% of all cases; small C-terminal deletions account for approximately 10%; and large deletions, 8% to 10%. MECP2 variant type is associated with disease severity. Whole duplications of the MECP2 gene have been associated with a severe X-linked intellectual disability with progressive spasticity, no or poor speech acquisition, and acquired microcephaly. Additionally, the pattern of X-chromosome inactivation influences the severity of the clinical disease females.

Because the spectrum of clinical phenotypes is broad, to facilitate genotype-phenotype correlation analyses, the International Rett Syndrome Association has established a locus-specific MECP2 variation database (RettBASE) and a phenotype database (InterRett).

Approximately 99.5% of cases of RTT are sporadic, resulting from a de novo variant, which arises almost exclusively on the paternally derived X chromosome. The remaining 0.5% of cases are familial and usually explained by germline mosaicism or favorably skewed X-chromosome inactivation in the carrier mother that results in her being unaffected or only slightly affected (mild intellectual disability). In the case of a carrier mother, the recurrence risk of RTT is 50%. If a variant is not identified in leukocytes of the mother, the risk to a sibling of the proband is below 0.5% (because germline mosaicism in either parent cannot be excluded).

Identification of a variant in MECP2 does not necessarily equate to a diagnosis of RTT. Rare cases of MECP2 variants also have been reported in other clinical phenotypes, including individuals with an Angelman-like picture, nonsyndromic X-linked intellectual disability, PPM-X syndrome (an X-linked genetic disorder characterized by psychotic disorders [most commonly bipolar disorder], parkinsonism, and intellectual disability), autism, and neonatal encephalopathy. Recent studies have revealed that different classes of genetic variants in MECP2 result in variable clinical phenotypes and overlap with other neurodevelopmental disorders.

A proportion of patients with a clinical diagnosis of RTT do not appear to have pathogenic variants in the MECP2 gene. Two other genes (CDKL5, FOXG1) have been shown to be associated with atypical variants.
Genetic Testing for Rett Syndrome

Policy # 00369
Original Effective Date: 11/20/2013
Current Effective Date: 01/09/2023

FDA or Other Governmental Regulatory Approval
U.S. Food and Drug Administration (FDA)
Clinical laboratories may develop and validate tests in-house and market them as a laboratory service; laboratory-developed tests must meet the general regulatory standards of the Clinical Laboratory Improvement Amendments (CLIA). Genetic testing for Rett syndrome is available under the auspices of the CLIA. Laboratories that offer laboratory-developed tests must be licensed by the CLIA for high-complexity testing. To date, the U.S. Food and Drug Administration (FDA) has chosen not to require any regulatory review of this test.

Rationale/Source
This medical policy was developed through consideration of peer-reviewed medical literature generally recognized by the relevant medical community, U.S. Food and Drug Administration approval status, nationally accepted standards of medical practice and accepted standards of medical practice in this community, technology evaluation centers, reference to federal regulations, other plan medical policies, and accredited national guidelines.

Description
Rett syndrome (RTT), a neurodevelopmental disorder, is usually caused by pathogenic variants in the methyl-CpG-binding protein 2 (MECP2) gene. Genetic testing is available to determine whether a pathogenic variant exists in RTT-associated genes (eg, MECP2, FOXG1, or CDLK5) in an individual with clinical features of RTT or an individual's family member.

Summary of Evidence
For individuals who have signs and/or symptoms of RTT who receive genetic testing for RTT-associated genes, the evidence includes case series and prospective cohort studies. Relevant outcomes are test accuracy and validity, other test performance measures, symptoms, health status measures, and quality of life. MECP2 variants are found in most patients with RTT, particularly in those who present with classic clinical features of RTT. The diagnostic accuracy of genetic testing for RTT cannot be determined with absolute certainty given variable clinical presentations of typical versus atypical RTT, but testing appears to have high sensitivity and specificity. Genetic testing has clinical utility when signs and symptoms of RTT are present to establish a specific genetic diagnosis. Identification of a specific class or type of pathogenic variant may alter some aspects of management and may eliminate or necessitate surveillance for different clinical manifestations of the disease. The
Genetic Testing for Rett Syndrome

Policy #   00369
Original Effective Date:  11/20/2013
Current Effective Date:  01/09/2023

evidence is sufficient to determine that the technology results in an improvement in the net health outcome.

For individuals who are asymptomatic sisters of an individual with RTT who receive targeted genetic testing for a known familial RTT-associated variant, the evidence includes case series and prospective cohort studies. Relevant outcomes are test accuracy and validity, other test performance measures, changes in reproductive decision making, symptoms, and symptoms. Targeted familial variant testing of asymptomatic sisters can eliminate or necessitate surveillance given the variability of clinical presentation in girls due to X-chromosome inactivation and clinical severity based on the type of pathogenic variant present. In sisters of reproductive age, determination of carrier status can eliminate or necessitate prenatal testing and inform reproductive decision making. The evidence is sufficient to determine that the technology results in an improvement in the net health outcome.

For individuals who are females with a child with RTT who are considering future childbearing who receive targeted genetic testing for a known familial RTT-associated variant, the evidence includes cases series and prospective cohort studies. Relevant outcomes are test accuracy and validity, other test performance measures, and changes in reproductive decision making. Targeted familial variant testing of a woman with a child with RTT to determine carrier status may inform prenatal testing and reproductive decision making. In the rare situation where the mother carries a pathogenic variant, all future offspring have a 50% of being affected, with males typically presenting with more severe disease. The evidence is sufficient to determine that the technology results in an improvement in the net health outcome.

**Supplemental Information**

**Clinical Input From Physician Specialty Societies and Academic Medical Centers**

While the various physician specialty societies and academic medical centers may collaborate with and make recommendations during this process, through the provision of appropriate reviewers, input received does not represent an endorsement or position statement by the physician specialty societies or academic medical centers, unless otherwise noted.

In response to requests, input on the use of variant testing for Rett syndrome (RTT) was received from 2 specialty medical societies (3 reviewers) and 3 academic medical centers, for a total of 6 reviewers, while this policy was under review in 2012. There was consensus or near consensus
Genetic Testing for Rett Syndrome

Policy #  00369
Original Effective Date:  11/20/2013
Current Effective Date:  01/09/2023

supporting the use of variant testing for the diagnosis of RTT in a girl in whom the clinical
differential diagnosis includes RTT, especially when clinical diagnosis is uncertain. Support for
testing sisters of individuals with RTT and prenatal screening was mixed.

Practice Guidelines and Position Statements
Guidelines or position statements will be considered for inclusion in ‘Supplemental Information’ if
they were issued by, or jointly by, a US professional society, an international society with US
representation, or National Institute for Health and Care Excellence (NICE). Priority will be given
to guidelines that are informed by a systematic review, include strength of evidence ratings, and
include a description of management of conflict of interest.

American Academy of Neurology and Child Neurology Society
In 2011, the American Academy of Neurology and the Child Neurology Society issued an evidence
report on genetic and metabolic testing of children with global developmental delay. The 2 societies
recommended considering methyl-CpG-binding protein 2 (MECP2) genetic testing for all girls with
unexplained moderate-to-severe developmental delay.

American Academy of Pediatrics
In 2007, the American Academy of Pediatrics (AAP) issued a policy statement (reaffirmed in 2014
and 2019) recommending MECP2 testing to confirm a diagnosis of suspected Rett syndrome (RTT),
especially when the diagnosis was unclear from symptoms alone.

In 2020, the AAP published Clinical Report Guidance on the identification, evaluation, and
management of children with autism spectrum disorder which stated that "if patient is a girl, consider
evaluation for Rett syndrome, MECP2 testing."

Neither the American Academy of Neurology nor the American Academy of Pediatrics has provided
recommendations on when to use CDKL5 or FOXG1 testing.

American College of Medical Genetics and Genomics
In 2013, the American College of Medical Genetics and Genomics revised its evidence-based
guidelines for clinical genetics evaluation of autism spectrum disorders. Testing for MECP2 genetic
variants was recommended as part of the diagnostic workup of females who present with an autistic
phenotype. Routine MECP2 testing in males with autism spectrum disorders was not recommended.
Genetic Testing for Rett Syndrome

Policy #  00369
Original Effective Date:  11/20/2013
Current Effective Date:  01/09/2023

U.S. Preventive Services Task Force Recommendations
Not applicable.

Medicare National Coverage
There is no national coverage determination. In the absence of a national coverage determination, coverage decisions are left to the discretion of local Medicare carriers.

Ongoing and Unpublished Clinical Trials
Some currently unpublished trials that might influence this review are listed in Table 1.

Table 1. Summary of Key Trials

<table>
<thead>
<tr>
<th>NCT No.</th>
<th>Trial Name</th>
<th>Planned Enrollment</th>
<th>Completion Date</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Ongoing</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>NCT02171104</td>
<td>MT2013-31: Allogeneic Hematopoietic Cell Transplantation for Inherited Metabolic Disorders and Severe Osteopetrosis Following Conditioning With Busulfan (Therapeutic Drug Monitoring), Fludarabine +/- ATG</td>
<td>100</td>
<td>Dec 2022</td>
</tr>
<tr>
<td><strong>Unpublished</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>NCT02153723</td>
<td>Pharmacological Treatment of Rett Syndrome With Glatiramer Acetate (Copaxone)</td>
<td>20</td>
<td>Jan 2016 (updated 11/05/2018)</td>
</tr>
<tr>
<td>NCT01777542</td>
<td>Pharmacological Treatment of Rett Syndrome by Stimulation of Synaptic Maturation With Recombinant Human IGF-1(Mecasermin [rDNA] Injection)</td>
<td>30</td>
<td>Nov 2016 (updated 03/26/2018)</td>
</tr>
<tr>
<td>NCT01520363</td>
<td>Placebo Controlled Trial of Dextromethorphan in Rett Syndrome</td>
<td>60</td>
<td>Oct 2016 (updated 12/04/2018)</td>
</tr>
</tbody>
</table>
Genetic Testing for Rett Syndrome

Policy # 00369
Original Effective Date: 11/20/2013
Current Effective Date: 01/09/2023

NCT: national clinical trial.

References
Genetic Testing for Rett Syndrome

Policy # 00369
Original Effective Date: 11/20/2013
Current Effective Date: 01/09/2023


Genetic Testing for Rett Syndrome

Policy #  00369
Original Effective Date:  11/20/2013
Current Effective Date:  01/09/2023


Policy History
Original Effective Date:  11/20/2013
Current Effective Date:  01/09/2023
11/07/2013  Medical Policy Committee review
12/04/2014  Medical Policy Committee review
08/03/2015  Coding update: ICD10 Diagnosis code section added; ICD9 Procedure code section removed.
12/03/2015  Medical Policy Committee review
12/16/2015  Medical Policy Implementation Committee approval. Coverage eligibility unchanged.
12/01/2016  Medical Policy Committee review
01/01/2017  Coding update: Removing ICD-9 Diagnosis Codes
12/07/2017  Medical Policy Committee review
12/20/2017  Medical Policy Implementation Committee approval. The policy is revised with updated genetics nomenclature. “Mutations” changed to “variants” in policy
Genetic Testing for Rett Syndrome

Policy # 00369
Original Effective Date: 11/20/2013
Current Effective Date: 01/09/2023

Statements. Policy rewritten with new PICO statements for indications 2 and 3 to limit populations to sisters of child with Rett syndrome (indication 2) or females with a child with Rett syndrome (indication 3) with the intervention revised to “targeted genetic testing for a known familial variant.” Policy statements updated to define “genetic testing for Rett syndrome–associated genes (e.g., MECP2, FOXG1, or CDKL5)”; Removed “female” requirement of child for testing; Added 2 new medical necessity statements for “targeted genetic testing for a known familial variant” in a sister of a child with Rett syndrome or a female with a child with Rett syndrome.

12/06/2018 Medical Policy Committee review
12/19/2018 Medical Policy Implementation Committee approval. Edited the investigational statement as follows: added examples of associated genes for Rett syndrome; added “routine” to describe carrier testing; added “persons with negative family history” to routine carrier testing.
12/05/2019 Medical Policy Committee review
12/03/2020 Medical Policy Committee review
12/02/2021 Medical Policy Committee review
02/15/2022 Coding update
12/01/2022 Medical Policy Committee review
12/14/2022 Medical Policy Implementation Committee approval. Coverage eligibility unchanged.

Next Scheduled Review Date: 12/2023

Coding
The five character codes included in the Blue Cross Blue Shield of Louisiana Medical Policy Coverage Guidelines are obtained from Current Procedural Terminology (CPT®), copyright 2021 by the American Medical Association (AMA). CPT is developed by the AMA as a listing of

©2022 Blue Cross and Blue Shield of Louisiana

Blue Cross and Blue Shield of Louisiana is an independent licensee of the Blue Cross and Blue Shield Association and incorporated as Louisiana Health Service & Indemnity Company.

No part of this publication may be reproduced, stored in a retrieval system, or transmitted, in any form or by any means, electronic, mechanical, photocopying, or otherwise, without permission from Blue Cross and Blue Shield of Louisiana.
Genetic Testing for Rett Syndrome

Policy # 00369
Original Effective Date: 11/20/2013
Current Effective Date: 01/09/2023

descriptive terms and five character identifying codes and modifiers for reporting medical services and procedures performed by physician.

The responsibility for the content of Blue Cross Blue Shield of Louisiana Medical Policy Coverage Guidelines is with Blue Cross and Blue Shield of Louisiana and no endorsement by the AMA is intended or should be implied. The AMA disclaims responsibility for any consequences or liability attributable or related to any use, nonuse or interpretation of information contained in Blue Cross Blue Shield of Louisiana Medical Policy Coverage Guidelines. Fee schedules, relative value units, conversion factors and/or related components are not assigned by the AMA, are not part of CPT, and the AMA is not recommending their use. The AMA does not directly or indirectly practice medicine or dispense medical services. The AMA assumes no liability for data contained or not contained herein. Any use of CPT outside of Blue Cross Blue Shield of Louisiana Medical Policy Coverage Guidelines should refer to the most current Current Procedural Terminology which contains the complete and most current listing of CPT codes and descriptive terms. Applicable FARS/DFARS apply.

CPT is a registered trademark of the American Medical Association.

Codes used to identify services associated with this policy may include (but may not be limited to) the following:

<table>
<thead>
<tr>
<th>Code Type</th>
<th>Code</th>
</tr>
</thead>
<tbody>
<tr>
<td>CPT</td>
<td>0234U, 81302, 81303, 81304, 81404, 81406</td>
</tr>
<tr>
<td></td>
<td>Delete codes effective 04/01/2022: 81470, 81471</td>
</tr>
<tr>
<td>HCPCS</td>
<td>No codes</td>
</tr>
<tr>
<td>ICD-10 Diagnosis</td>
<td>F84.2, G31.81, G31.82</td>
</tr>
</tbody>
</table>

*Investigational – A medical treatment, procedure, drug, device, or biological product is Investigational if the effectiveness has not been clearly tested and it has not been incorporated into standard medical practice. Any determination we make that a medical treatment, procedure, drug, device, or biological product is Investigational will be based on a consideration of the following:
  A. Whether the medical treatment, procedure, drug, device, or biological product can be lawfully marketed without approval of the U.S. Food and Drug Administration (FDA) and

©2022 Blue Cross and Blue Shield of Louisiana

Blue Cross and Blue Shield of Louisiana is an independent licensee of the Blue Cross and Blue Shield Association and incorporated as Louisiana Health Service & Indemnity Company.

No part of this publication may be reproduced, stored in a retrieval system, or transmitted, in any form or by any means, electronic, mechanical, photocopying, or otherwise, without permission from Blue Cross and Blue Shield of Louisiana.
Genetic Testing for Rett Syndrome

Policy # 00369
Original Effective Date: 11/20/2013
Current Effective Date: 01/09/2023

whether such approval has been granted at the time the medical treatment, procedure, drug, device, or biological product is sought to be furnished; or
B. Whether the medical treatment, procedure, drug, device, or biological product requires further studies or clinical trials to determine its maximum tolerated dose, toxicity, safety, effectiveness, or effectiveness as compared with the standard means of treatment or diagnosis, must improve health outcomes, according to the consensus of opinion among experts as shown by reliable evidence, including:
   1. Consultation with technology evaluation center(s);
   2. Credible scientific evidence published in peer-reviewed medical literature generally recognized by the relevant medical community; or
   3. Reference to federal regulations.

**Medically Necessary (or “Medical Necessity”) - Health care services, treatment, procedures, equipment, drugs, devices, items or supplies that a Provider, exercising prudent clinical judgment, would provide to a patient for the purpose of preventing, evaluating, diagnosing or treating an illness, injury, disease or its symptoms, and that are:
   A. In accordance with nationally accepted standards of medical practice;
   B. Clinically appropriate, in terms of type, frequency, extent, level of care, site and duration, and considered effective for the patient's illness, injury or disease; and
   C. Not primarily for the personal comfort or convenience of the patient, physician or other health care provider, and not more costly than an alternative service or sequence of services at least as likely to produce equivalent therapeutic or diagnostic results as to the diagnosis or treatment of that patient's illness, injury or disease.

For these purposes, “nationally accepted standards of medical practice” means standards that are based on credible scientific evidence published in peer-reviewed medical literature generally recognized by the relevant medical community, Physician Specialty Society recommendations and the views of Physicians practicing in relevant clinical areas and any other relevant factors.

‡ Indicated trademarks are the registered trademarks of their respective owners.

NOTICE: If the Patient’s health insurance contract contains language that differs from the BCBSLA Medical Policy definition noted above, the definition in the health insurance contract will be relied upon for specific coverage determinations.
Genetic Testing for Rett Syndrome

Policy # 00369
Original Effective Date: 11/20/2013
Current Effective Date: 01/09/2023

NOTICE: Medical Policies are scientific based opinions, provided solely for coverage and informational purposes. Medical Policies should not be construed to suggest that the Company recommends, advocates, requires, encourages, or discourages any particular treatment, procedure, or service, or any particular course of treatment, procedure, or service.