DNA Based Testing for Adolescent Idiopathic Scoliosis

**Description of Procedure or Service**

Adolescent idiopathic scoliosis (AIS) is the most common pediatric spinal deformity affecting 1 to 3% of adolescents. This disease, of unknown etiology, occurs in otherwise healthy children with the onset of, and highly correlated with, the adolescent growth spurt. The vertebrae become misaligned such that the spine deviates from the midline laterally and rotates axially. Deviation can occur anteriorly (a lordotic deviation), posteriorly (a kyphotic deviation) or laterally. Although AIS affects females and males in a nearly 1:1 ratio, progression to severe deformity occurs more often in females. Because the disease can have rapid onset and produce considerable morbidity, school screenings have been recommended. However, screening remains somewhat controversial, with conflicting guidelines supporting and not supporting this practice.

The familial nature of this disease was noted as early as 1968. About one-quarter of patients report a positive family history of disease and twin studies have consistently supported shared genetic factors. Genome-wide linkage studies have reported multiple chromosomal regions of interest, often not replicated. DNA testing to predict the risk of progression of AIS is a novel development in the field of genetics.

**Regulatory Status**

The ScoliScore™AIS prognostic DNA-based test (originally developed by Axial Biotech, Salt Lake City, UT; test rights acquired by Transgenomic in 2013) has not been approved or cleared by the U.S Food and Drug Administration (FDA), but is being offered as a laboratory-developed test. The laboratory performing this test is accredited by the Centers for Medicare and Medicaid (CMS) under the Clinical Laboratory Improvement Amendments of 1988 (CLIA).

As of December 2016, the Transgenomic website did not include any information about the ScoliScore test.

***Note: This Medical Policy is complex and technical. For questions concerning the technical language and/or specific clinical indications for its use, please consult your physician.***

**Policy**

DNA based prognostic testing for adolescent idiopathic scoliosis is considered investigational. BCBSNC does not provide coverage for investigational services or procedures.

**Benefits Application**

This medical policy relates only to the services or supplies described herein. Please refer to the Member's Benefit Booklet for availability of benefits. Member's benefits may vary according to benefit
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design; therefore member benefit language should be reviewed before applying the terms of this medical policy.

When DNA Based testing for Adolescent Idiopathic Scoliosis is covered

Not Applicable

When DNA Based Testing for Adolescent Idiopathic Scoliosis is not covered

DNA based prognostic testing for adolescent idiopathic scoliosis is considered investigational. BCBSNC does not cover investigational services or procedures.

Policy Guidelines

Adolescent idiopathic scoliosis is a disease of unknown etiology that causes mild to severe spinal deformity in approximately 1% to 3% of adolescents. While there is controversy about the value of both screening and treatment, patients once diagnosed are frequently closely followed. In cases with significant progression of curvature, both medical (bracing) and surgical (spinal fusion) interventions are considered. The ScoliScore AIS prognostic DNA-based test uses an algorithm incorporating results of testing for 53 single-nucleotide variants (SNVs), along with the patient’s presenting spinal curve (Cobb angle), to generate a risk score (range, 1-200), which can be used qualitatively or quantitatively to predict the likelihood of spinal curve progression.

For individuals with AIS who receive clinical management with prognostic testing with an algorithm incorporating SNV-based testing, the evidence includes cross-sectional studies reporting on the clinical validity of the ScoliScore test, along with cross-sectional studies reporting on the association between SNVs in various genes and scoliosis progression. Relevant outcomes are symptoms, morbid events, and change in disease status. A single study on the clinical validity for the ScoliScore AIS prognostic DNA-based test has reported a high negative predictive value for ruling out the possibility of progression to severe curvature in a population with a low baseline likelihood of progression. It is not clear if the increase in predictive accuracy provided by testing is statistically or clinically meaningful. Other genetic studies have not demonstrated significant associations between the SNVs used in the ScoliScore and scoliosis progression. Studies have identified additional SNVs that may be associated with AIS severity, but these associations have not been reliably replicated. The clinical validity of DNA-based testing (either through testing of individual SNVs or through an algorithm incorporating SNV results) for predicting scoliosis progression in patients with AIS has not been established. There is no direct evidence demonstrating that use of this test results in changes in management that improve outcomes. The value of early identification and intervention(s) for people at risk for progression of disease and whether laboratory testing improves disease identification beyond clinical evaluation is unknown. The evidence is insufficient to determine the effects of the technology on health outcomes.

In 2004, the U.S. Preventive Services Task Force (USPSTF) recommended against the routine screening of asymptomatic adolescents for idiopathic scoliosis (Grade D Recommendation). No USPSTF recommendations for DNA-based testing for adolescent idiopathic scoliosis were identified.

Billing/Coding/Physician Documentation Information

This policy may apply to the following codes. Inclusion of a code in this section does not guarantee that it will be reimbursed. For further information on reimbursement guidelines, please see Administrative Policies on the Blue Cross Blue Shield of North Carolina web site at www.bcbsnc.com. They are listed in the Category Search on the Medical Policy search page.
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Applicable service codes: 0004M, G0452, 81479

BCBSNC may request medical records for determination of medical necessity. When medical records are requested, letters of support and/or explanation are often useful, but are not sufficient documentation unless all specific information needed to make a medical necessity determination is included.

Scientific Background and Reference Sources


Specialty Matched Consultant Advisory Panel review 2/2012

Specialty Matched Consultant Advisory Panel review 2/2013

Specialty Matched Consultant Advisory Panel review 2/2014

Medical Director review 2/2014


Specialty Matched Consultant Advisory Panel review 2/2015


Specialty Matched Consultant Advisory Panel review 2/2017

Policy Implementation/Update Information

10/11/11 New policy developed. DNA based testing for adolescent idiopathic scoliosis is not covered. It is considered investigational. BCBSNC does not cover investigational services or procedures. Medical Director review 8/2011. (mco)
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3/20/12  Specialty Matched Consultant Advisory Panel review 2/2012. No changes to policy statement. (mco)

10/30/12  References updated. No changes to Policy Statement. (mco)

1/1/13    Deleted the following statement from the Billing/Coding section: “There is no specific CPT code for this test. A series of molecular diagnostic codes such as 83891, 83898, 83903, 83012 would likely be used.” Added code 81599, G0452 to Billing/Coding section. (mco)


7/1/13    Deleted unlisted CPT code 81599 and added CPT code 0004M to Billing/Coding section. (mco)

8/27/13   References update. No changes to Policy Statement. (mco)


8/26/14   Policy Guidelines and References updated. No changes to Policy Statements. (mco)


9/1/15    Reference added. (sk)

4/1/16    Specialty Matched Consultant Advisory Panel review 2/24/2016. (sk)


Medical policy is not an authorization, certification, explanation of benefits or a contract. Benefits and eligibility are determined before medical guidelines and payment guidelines are applied. Benefits are determined by the group contract and subscriber certificate that is in effect at the time services are rendered. This document is solely provided for informational purposes only and is based on research of current medical literature and review of common medical practices in the treatment and diagnosis of disease. Medical practices and knowledge are constantly changing and BCBSNC reserves the right to review and revise its medical policies periodically.