

Overview

Useful For

Diagnosing open neural tube defects and, to a lesser degree, ventral wall defects

Special Instructions

- [Second Trimester Maternal Screening Alpha-Fetoprotein \(AFP\)/Quad Screen Patient Information](#)
- [Biochemical Genetics Patient Information](#)

Method Name

PolyacrylamideElectrophoresis

NY State Available

Yes

Specimen

Specimen Type

Amniotic Fld

Additional Testing Requirements

If chromosome studies are also requested, see CHRAF / Chromosome Analysis, Amniotic Fluid for specimen requirements. When requested with chromosome analysis, the specimen cannot be frozen.

Necessary Information

Gestational age at amniocentesis is required.

Specimen Required

Container/Tube: Amniotic fluid container

Specimen Volume: 1 mL

Collection Instructions: A specimen from the 14 to 18 week gestational period of pregnancy is preferred. Amniotic fluid from the 14 to 21 week gestational period is acceptable.

Forms

1. [Second Trimester Maternal Screening Alpha-Fetoprotein \(AFP\)/QUAD Screen Patient Information \(T595\)](#) is required; see Special Instructions.
2. [Biochemical Genetics Patient Information](#) (T602) in Special Instructions.

Specimen Minimum Volume

0.3 mL

Reject Due To

Gross hemolysis	OK
Gross icterus	OK

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Amniotic Fld	Refrigerated (preferred)	365 days	
	Frozen	365 days	
	Ambient	14 days	

Clinical and Interpretive

Clinical Information

Neural tube defects (NTD) are a type of birth defect involving openings along the brain and spine. They develop in the early embryonic period when the neural tube fails to completely close. NTD can vary widely in severity. Anencephaly represents the most severe type of NTD, which occurs when the cranial end fails to develop properly, resulting in an absence of the forebrain, the area of the skull that covers the brain, and the skin. Most infants with anencephaly are stillborn or die shortly after birth. NTD along the spine are referred to as spina bifida. Individuals with spina bifida may experience hydrocephalus, urinary and bowel dysfunction, club foot, lower body weakness, and loss of feeling or paralysis. Severity varies depending upon whether the NTD is covered by skin, whether herniation of the meninges and spinal cord are present, and the location of the lesion. NTD not covered by skin are referred to as open NTD and are typically more severe than closed NTD. Likewise those presenting with herniation and higher on the spinal column are typically more severe.

Most NTD occur as isolated birth defects with an incidence of approximately 1 to 2 in 1,000 live births in the United States. Rates vary by geographic region with lower rates being observed in the North and West than the South and East. A fetus is at higher risk when the pregnancy is complicated by maternal diabetes, exposed to certain anticonvulsants, or there is a family history of NTD. Studies have shown a dramatic decrease in risk as a result of maternal dietary supplementation with folic acid. The March of Dimes currently recommends that all women of childbearing age take 400 mcg of folic acid daily, increasing the amount to 600 mcg/day during pregnancy. For women who have had a prior pregnancy affected by an NTD, the recommended dose is at least 4000 mcg/day starting at least 1 month preconception and continuing through the first trimester.

When an NTD is suspected based upon maternal serum alpha-fetoprotein (AFP) screening results or diagnosed via ultrasound, analysis of AFP and acetylcholinesterase (AChE) in amniotic fluid are useful diagnostic tools. AChE is primarily active in the central nervous system with small amounts of enzyme found in erythrocytes, skeletal muscle, and fetal serum. Normal amniotic fluid does not contain AChE, unless contributed by the fetus as a result of an open NTD.

Reference Values

Negative (reported as negative [normal] or positive [abnormal] for inhibitable acetylcholinesterase)

Reference values were established in conjunction with alpha-fetoprotein testing and include only amniotic fluids from pregnancies between 14 and 21 weeks gestation.

Interpretation

The presence of acetylcholinesterase in amniotic fluid is consistent with open neural tube defects and, to a lesser degree, ventral wall defects.

Cautions

False-positive acetylcholinesterase (AChE) results may occur when blood is present in the amniotic fluid specimen or due to contamination from fetal calf serum.

Clinical Reference

1. Wilson RD, Audibert F, Brock JA, et al: Prenatal screening, diagnosis, and pregnancy management of fetal neural tube defects. J Obstet Gynaecol Can 2014 Oct;36(10):927-939
2. Palomaki GE, Bupp C, Gregg AR, et al: Laboratory screening and diagnosis of open neural tube defects, 2019 revision: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genet Med 2020 Mar;22(3):462-474

Performance

Method Description

Acetylcholinesterase (AChE) and pseudocholinesterase (PChE) catalyze the deesterification of acetylthiocholine iodide to form a white copper thiocholine precipitate. The white precipitate is acted upon by dithiooxamide to provide a dark-green color. If 1,5 bis (4-allyldimethylammoniumphenyl) pentane-3-1 dibromide (BW284C51) is present, it will inhibit the action of AChE on acetylthiocholine iodide but will not inhibit the activity of PChE. AChE and PChE are separated by polyacrylamide gel electrophoresis and then reacted with the inhibitor and substrate while remaining in the gel.(Barlow RD, Cuckle HS, Wald NJ: A simple method for amniotic fluid gel-acetylcholinesterase determination, suitable for routine use in the antenatal diagnosis of open neural tube defects. Clin Chim Acta 1982;119:137-142); Cowan T, Pasquali M: Laboratory Investigations of Inborn Errors of Metabolism. In Pediatric Endocrinology and Inborn Errors of Metabolism. Second Edition. Edited by K Sarafoglou, GF Hoffman, KS Roth. 2017. pp 1139-1158)

PDF Report

No

Day(s) Performed

Thursday

Report Available

8 to 15 days

Specimen Retention Time

60 days

Performing Laboratory Location

Rochester

Fees and Codes

Fees

- Authorized users can sign in to [Test Prices](#) for detailed fee information.
- Clients without access to Test Prices can contact [Customer Service](#) 24 hours a day, seven days a week.
- Prospective clients should contact their Regional Manager. For assistance, contact [Customer Service](#).

Test Classification

This test was developed, and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

82013

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
ACHE_	Acetylcholinesterase, AF	30106-9

Result ID	Test Result Name	Result LOINC Value
9287	Acetylcholinesterase, AF	30106-9
GACHE	Gestational Age (ACHE)	18185-9