MAYO CLINIC LABORATORIES

# Test Definition: ACHE_

Acetylcholinesterase, Amniotic Fluid

## Overview

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

### Special Instructions
- Second Trimester Maternal Screening Alpha-Fetoprotein / Quad Screen Patient Information

### Method Name
Polyacrylamide Electrophoresis

### 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
1. **Gestational age at amniocentesis is required**.
2. If not ordering electronically, provide gestational age on Second Trimester Maternal Screening Alpha-Fetoprotein / Quad Screen Patient Information (T595) and send with specimen.

### Specimen Required
**Container/Tube:** Amniotic fluid container
**Specimen Volume:** 1 mL
**Collection Instructions:** Specimen must be collected between 14 to 21 weeks gestation; 14 to 18 weeks is preferred.

### Forms
Second Trimester Maternal Screening Alpha-Fetoprotein / Quad Screen Patient Information (T595)

### Specimen Minimum Volume
0.3 mL

### Reject Due To

| | |
|---|---|
| Gross | OK |

| | |
|---|---|
| hemolysis | |
| Gross icterus | OK |

### Specimen Stability Information

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

## Clinical & 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 close completely. 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 1000 live births in the United States. Rates vary by geographic region with lower rates being observed in the North and West than in 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 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 red blood cells, 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 results may occur when blood is present in the amniotic fluid specimen or due to contamination from fetal calf serum.

### Clinical Reference

1. Douglas Wilson R, Van Mieghem T, Langlois S, Church P. Guideline No. 410: Prevention, Screening, Diagnosis, and Pregnancy Management for Fetal Neural Tube Defects. J Obstet Gynaecol Can. 2021;43(1):124-139.e8. doi:10.1016/j.jogc.2020.11.003

2. Palomaki GE, Bupp C, Gregg AR, Norton ME, Oglesbee D. 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;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 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: Sarafoglou K, Hoffman GF, Roth KS, eds. Pediatric Endocrinology and Inborn Errors of Metabolism. 2nd ed. McGraw Hill; 2017:1139-1158)

### PDF Report

No

### Day(s) Performed

Tuesday, Thursday

### Report Available

3 to 7 days

### Specimen Retention Time

60 days

### Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

## Fees & 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 account representative. 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. It 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 |