MAYO CLINIC LABORATORIES

Test Definition: YMCRO

Y Chromosome Microdeletions, Molecular Detection, Varies

Overview

Useful For

Evaluating men with azoospermia, severe oligozoospermia, or otherwise unexplained male factor infertility

Genetics Test Information

Tests for the presence of microdeletions in the *AZFa*, *AZFb*, and *AZFc* regions of the Y chromosome.

Special Instructions

- Molecular Genetics: Congenital Inherited Diseases Patient Information
- Informed Consent for Genetic Testing
- Informed Consent for Genetic Testing (Spanish)

Method Name

Polymerase Chain Reaction (PCR)

NY State Available

Yes

Specimen

Specimen Type

Varies

Specimen Required

Patient Preparation: A previous bone marrow transplant from an allogeneic donor will interfere with testing. For instructions for testing patients who have received a bone marrow transplant, call 800-533-1710.
Specimen Type: Whole blood
Container/Tube:
Preferred: Lavender top (EDTA) or yellow top (ACD)
Acceptable: None
Specimen Volume: 3 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. **Do not aliquot**.
Additional Information:
1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed.
2. To ensure minimum volume and concentration of DNA is met, the preferred volume of blood must be submitted. Testing may be canceled if DNA requirements are inadequate.

Forms

1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing-Spanish (T826)

2. Molecular Genetics: Congenital Inherited Diseases Patient Information (T521)

Specimen Minimum Volume

See Specimen Required

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Ambient (preferred)		
	Refrigerated		
	Frozen		

Clinical & Interpretive

Clinical Information

Yq microdeletions involving some or all azoospermic factor (AZF) regions, are identified in approximately 3% of infertile men. Yq microdeletions are also the most frequently identified cause of spermatogenic failure in chromosomally normal men with nonobstructive azoospermia (3%-15%) or severe oligospermia (6%-10%). The relative frequency of Yq microdeletions makes the evaluation for them an important aspect of the diagnostic work up in infertile men, especially those with azoospermia or severe oligospermia.

Most cases of Yq microdeletions occur *de novo*, and due to the consequential infertile phenotype, they are typically not transmitted. However, in cases where assisted reproductive technology (eg, testicular sperm extraction followed by intracytoplasmic sperm injection) is used to achieve viable pregnancy, all male offspring born to a microdeletion carrier will carry the deletion and may be infertile.

Men testing positive for 1 or more microdeletions who are enrolled in an in vitro fertilization treatment program may wish to consider alternative options to intracytoplasmic sperm injection (eg, donor sperm), and consultation with an experienced reproductive endocrinologist and medical geneticist is recommended.

Most Y microdeletions are the result of homologous recombination between repeated sequence blocks. Testing for deletions involves investigating for the presence or absence of markers located within nonpolymorphic regions of the AZF region.

Reference Values

An interpretive report will be provided.

Interpretation

The interpretive report includes an overview of the findings as well as the associated clinical significance.

Cautions

This assay will not detect all causes of infertility or azoospermia. Therefore, the absence of a detectable microdeletion does not rule out the presence of other genetic or nongenetic factors that may be the cause of clinical findings.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in the interpretation of results may occur if information given is inaccurate or incomplete.

Rare variants (ie, polymorphisms) exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

In rare cases, DNA alterations of undetermined significance may be identified.

A genetic consultation is recommended for all patients undergoing this testing. Additional consultation with a reproductive endocrinologist/urologist to discuss reproductive options is recommended when a deletion is detected.

Supportive Data

Validation studies done at Mayo Clinic on a series of known fertile and infertile specimens provided the following results. Of 111 DNA specimens from known fertile men, 110 gave unequivocal negative results demonstrating clinical specificity of 99%. A series of 19 specimens from females (negative controls) were all negative, as expected. In a small series of specimens from 4 men being treated for male factor infertility, no deletions were found (which may be expected given a reported prevalence of 7% for microdeletions in unselected male infertility patients). Seven specimens were mailed in from outside laboratories (3 of which were sent as part of an external quality assessment scheme organized by the European Academy of Andrology) with known deletions in either AZFa, AZFb, or AZFc. Using this assay, all were diagnosed correctly to give an analytical sensitivity of 100%.

Clinical Reference

1. Stahl PJ, Masson P, Mielnik A, Marean MB, Schlegel PN, Paduch DA. A decade of experience emphasizes that testing for Y microdeletions is essential in American men with azoospermia and severe oligozoospermia. Fertil Steril. 2010;94(5):1753-1756
2. Bhasin S. Approach to the infertile man. J Clin Endocrinol Metab. 2007;92(6):1995-2004
3. Fan Y, Silber SJ. Y Chromosome Infertility. In: Adam MP, Feldman J, Mirzaa GM, et al, eds. GeneReviews [Internet]. University of Washington, Seattle; 2002. Updated August 1, 2019. Accessed November 18, 2024. Available at www.ncbi.nlm.nih.gov/books/NBK1339/
4. Kim SY, Kim HJ, Lee BY, Park SY, Lee HS, Seo JT. Y Chromosome Microdeletions in Infertile Men with Non-obstructive Azoospermia and Severe Oligozoospermia. J Reprod Infertil. 2017;18(3):307-315

Performance

Method Description
Multiplex polymerase chain reaction and agarose gel electrophoresis are used to test DNA for the presence of microdeletions in the AZFa, AZFb, and AZFc regions of the Y chromosome.(Simoni M, Bakker E, Krausz C. EAA/EMQN best practice guidelines for molecular diagnosis of y-chromosomal microdeletions. State of the art 2004. Int J Androl. 2004;27[4]:240-249)

PDF Report
No

Day(s) Performed
Wednesday

Report Available
7 to 10 days

Specimen Retention Time
Whole blood: 2 weeks (if available); Extracted DNA: 3 months

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
81403-DAZ/SRY (deleted in azoospermia and sex determining region Y) (eg, male infertility), common deletions (eg, AZFa, AZFb, AZFc, AZFd)

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
YMCRO	Y Microdeletion	35456-3

Result ID	Test Result Name	Result LOINC® Value
53364	Result Summary	50397-9

53365	Result	82939-0
53366	Interpretation	69047-9
53367	Specimen	31208-2
53368	Source	31208-2
53369	Released By	18771-6