

Specimen Identity Testing

TO DETERMINE PATIENT ORIGIN FOR SAMPLES, TISSUES, OR TISSUE FRAGMENTS

Test Highlights

- Confirm identity of possible switched specimens
- Confirm floaters in tissue preparations

Clinical Background

- Specimen mix-up or switching is a constant concern in clinical testing.
- Specimen mix-up may occur at the time of collection or during the testing process.
- Floater tissue from a different patient may occur in tissue paraffin samples or on slides.
- Genetic analysis based on short-tandem repeats can show a match or mismatch to a reference sample.
- The same analysis can show that tissue fragments on the same slide are from the same or different individual.

Genetics

- Short tandem repeats (microsatellites) have tremendous variability between individuals.
- The probability that a random individual will have the identical genotype as the specimen in question with this assay is less than one in 10^{16} . Probabilities for ethnic backgrounds are as follows:
 - African Americans: one in 7.64×10^{17}
 - Caucasians: one in 2.00×10^{17}
 - U.S. Hispanics: one in 1.31×10^{17}
 - Native Americans: one in 2.76×10^{16}
- Probabilities for other ethnic backgrounds are not available.

Indications for Ordering

- Suspicion that a sample has been switched. A sample (blood or cheek swab) that can be used as a reference sample should accompany the test request.
- Suspicion that a tissue fragment on a slide(s) is from a different individual (floater).

Interpretation

- Match: The sample in question matches a reference sample at 16 of 16 markers. The probability that the sample comes from an individual different from the reference is less than one in 10^{16} .
- Excluded: The sample in question is excluded from being from the same individual as the reference sample.
- Probable match: Genetic instability was evident in the tumor. However, germline and tumor markers showed enough similarity to indicate a probable match.
- Floater: The tissue fragment in question comes from a different individual.
- Not a floater: The tissue fragment in question probably comes from the same individual.

Limitations

- Genetic instability in a tumor may complicate interpretation.
- ARUP prohibits use of this test for forensic testing.

Methodology

- PCR amplification of 16 STR markers followed by capillary electrophoresis
- STR profiler kit (AB) used

Reference

1. Williams TM, Weedn VW, Kamoun M. Identity assessment. *In: Tietz Textbook of Clinical Chemistry and Molecular Diagnostics*. 4th ed. Burtis CA, Ashwood ER, Bruns DE, Eds. Philadelphia, Elsevier Saunders, 2006, pp. 1545–6.

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Test Information

0051139 Specimen Identity Testing

For specific collection, transport, and testing information, refer to the ARUP Web site at www.aruplab.com.