ASSOCIATION FOR MOLECULAR PATHOLOGY
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AMP 2025 press materials available

Get the latest molecular diagnostics research news being presented at the Association for Molecular Pathology's annual meeting this week

ROCKVILLE, Md. — Press materials are now available for the **Association for Molecular Pathology 2025 Annual Meeting & Expo**. Top clinicians, scientists and educators in the field will gather at the meeting Nov. 11–15 in Boston.

Featured research findings include:

- Faster molecular test for dangerous regional **fungal infections**
- New test for detecting **Lyme disease** earlier
- Multiple breakthrough studies in **acute myeloid leukemia**
- **AI tools** for improving molecular diagnosis
- New test to identify cases of **rare movement disorder**
- Optical genome mapping to reveal causes **recurrent pregnancy loss**
- Using **shelved DNA samples** to understand the evolution of colorectal cancer

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New analysis technique provides new insight into old DNA samples

Researchers at the University of Chicago adapted techniques originally used to study ancient DNA from archaeological specimens to recover genetic information from old medical samples.

Why it matters:

- Traditional methods make analyzing DNA samples over 20 years old difficult
- New technique could track disease changes across decades
- May help doctors uncover root causes of previously unknown triggers of diseases

Key findings:

- Able to analyze DNA samples stored nearly 100 years ago
- Proved adaptability of archaeological genomics techniques
- Uncovered potential cause of increase of colon cancer cases in young adults

Read the full release.

Faster molecular test for dangerous regional fungal infections

Researchers at Indiana University Health have created a test that rapidly detects three serious U.S. fungal pathogens often misdiagnosed as other respiratory illnesses: Histoplasma, Blastomyces, and Coccidioides.

Why it matters:

- Current diagnostics rely on cultures that take weeks
- Early symptoms mimic pneumonia or viral infections
- No FDA-approved molecular tests currently exist

Key findings:

- Multiplex real-time PCR test achieved 100% sensitivity and specificity
- Detects fungal DNA directly from samples, bypassing lengthy culture
- Eliminates DNA extraction step to reduce lab time and biosafety risk

Read the full release.

Test shows promise for detecting Lyme disease earlier

Researchers at Dartmouth Hitchcock Medical Center have developed a droplet digital PCR (ddPCR) test that may detect Lyme disease more accurately and earlier than traditional methods — potentially avoiding years-long diagnostic delays that can lead to severe complications.

Why it matters:

- Lyme disease cases are increasing, especially in the Northeast
- Antibody tests can be false-negative early in infection
- Delayed diagnosis can lead to disabling joint, heart and neurological issues

Key findings:

- ddPCR assay detects *Borrelia burgdorferi* DNA directly
- Detection threshold as low as 5–10 bacterial cells
- Successfully diagnosed a patient with a four-year diagnostic delay who had negative antibody results but responded to antibiotics

Read the full release.

Multiple breakthrough studies in acute myeloid leukemia

- **Genetic mutations after transplant may predict AML relapse:** UC San Diego study tracked mutations before and after stem cell transplant in 74 patients. Persistent mutations in two genes strongly predicted relapse. The work could improve post-transplant monitoring and follow-up treatment decisions.

- **RNA-based test detects hidden AML gene fusions:** University of Michigan scientists evaluated 600 AML cases by integrating RNA fusion analysis into standard next-generation sequencing, finding gene fusions in 15% of patients and identifying 23 cryptic fusions missed by cytogenetics, including some that were clinically actionable.

- **Ultra-sensitive sequencing for residual AML:** Moffitt Cancer Center researchers validated a deep-sequencing test for *FLT3* mutations, which are common in AML and linked to higher relapse rate. It could be a powerful tool for detecting relapse earlier and guiding transplant decisions.

Read the tipsheet.

AI tools show major promise in improving molecular diagnosis

- **AI classifier achieves 93% accuracy in cancer diagnosis:** Researchers at The Hospital for Sick Children created a web-based AI platform to classify cancers using RNA-sequencing data — even when samples vary in preparation and storage.

- **AI enables early, noninvasive diagnosis of central nervous system tumors:** A team at Soonchunhyang University (South Korea) trained two AI models to diagnose tumors using cerebrospinal fluid circulating tumor DNA and MRI imaging — offering a potential alternative to invasive biopsies. The approach may allow earlier diagnosis and better surgical planning without requiring repeated brain or spinal biopsies.

- **AI-assisted karyotyping reveals complex chromosomal changes in leukemia:** At Wake Forest University School of Medicine, researchers used an AI-based karyotyping tool to analyze chromosomal abnormalities in GATA2 deficiency syndrome, a rare condition linked to blood cancers. This approach may provide clearer insight into disease trajectory and relapse risk in myeloid malignancies.

- **AI framework predicts tumor genomics from slide images:** Researchers at Augusta University developed a computational pipeline comparing AI models that predict genetic and transcriptomic tumor features from routine slide images, aiming to reduce reliance on expensive sequencing tests. Long-term, this approach may help deliver precision oncology insights directly from pathology slides, accelerating and broadening access to personalized cancer care.

Read the tipsheet.

New genetic test finds hidden cases of rare movement disorder

A Harvard/Brigham and Women's Hospital team developed a targeted DNA test for X-linked dystonia-parkinsonism, a rare inherited movement disorder primarily affecting men of Filipino descent.

Why it matters:

- Symptoms mimic Parkinson's disease, leading to frequent misdiagnosis
- Standard gene panels and exome sequencing often miss the mutation
- Early detection supports genetic counseling and care planning

Key findings:

- Able to analyze DNA samples
- Correctly identified all known carriers and three previously misdiagnosed cases
- Two patients had false-negative results on standard genetic tests

Read the full release.

Technique helps doctors identify possible causes of pregnancy loss

Two groups of researchers — one at Dartmouth–Hitchcock Medical Center and one at Queens University's Kingston Health Sciences Centre and the University of Ottawa — found that optical genome mapping could help uncover causes of recurrent pregnancy loss.

Why it matters:

- Pregnancy loss may occur in up to 25% of all pregnancies
- The cause of recurrent pregnancy loss is difficult to pinpoint
- May help doctors uncover previously unknown causes

Key findings:

- Found structural chromosome changes missed by traditional testing methods
- Identified possible cause of recurrent pregnancy loss that was previously overlooked
- Both studies provided a more detailed picture of genetic material

Read the tipsheet.

About AMP

The Association for Molecular Pathology was founded in 1995 to provide structure and leadership to the emerging field of molecular diagnostics. AMP's more than 3,100 members practice various disciplines of molecular diagnostics, including bioinformatics, infectious diseases, inherited conditions and oncology. Our members are pathologists, clinical laboratory directors, basic and translational scientists, technologists and trainees who practice in a variety of settings, including academic and community medical centers, government and industry. Through the efforts of its Board of Directors, Committees, Working Groups and Members, AMP is the primary resource for expertise, education and collaboration in one of the fastest-growing fields in healthcare. AMP members influence policy and regulation on the national and international levels, ultimately serving to advance innovation in the field and protect patient access to high-quality, appropriate testing. For more information, visit www.amp.org.

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