

For Immediate Release

**>>Signature Genomic Laboratories detects chromosome abnormalities in individuals with Pallister-Killian syndrome without invasive skin biopsy**

Spokane, Wash. –Geneticists at [Signature Genomic Laboratories](http://www.signaturegenomics.com) have demonstrated that microarray-based genetic testing can identify a rare genetic disorder using DNA from blood rather than the more-invasive skin biopsy routinely used for testing.

Pallister-Killian syndrome (PKS) is a rare genetic disorder characterized by mental retardation, seizures, streaks of hypo- or hyperpigmentation and coarse facial features. PKS results from the presence of four, rather than the normal two, copies of the short arm of chromosome 12 in some of the body's cells. The extra fragments of DNA usually cannot be identified in the cultured cells derived from blood used for conventional chromosome analysis, necessitating a painful skin biopsy for diagnosis. In their study, published in the May issue of the [\*American Journal of Medical Genetics\*](#), geneticists at Signature reported eight individuals referred for testing for unexplained mental retardation or developmental delay in whom microarray analysis of uncultured DNA from peripheral blood identified a two-copy gain of the short arm of chromosome 12. In all but one individual, traditional cytogenetic analysis using cultured cells could not identify the abnormality, a discrepancy the authors attribute to the inability of abnormal cells to compete with normal cells during the culturing process required for conventional chromosome analysis. The authors suggest that, because it does not require cultured cells for analysis, microarray analysis can better detect chromosome abnormalities that are not present in every cell.

“These results show that microarray analysis can circumvent some of the inherent technical limitations of traditional chromosome analysis in the identification of abnormalities that are not present in every cell,” said Dr. Lisa G. Shaffer, Ph.D., President and CEO of Signature and senior author of the study. “Furthermore, although Pallister-Killian syndrome is rare, individuals suspected to have the disorder can be tested using DNA derived from a simple blood draw rather than submit to a painful and invasive skin biopsy.”

**About Signature Genomic Laboratories, LLC**

Signature Genomic Laboratories is the leader in providing microarray-based chromosome analysis. Signature's worldwide client base includes clinical geneticists, neurologists, pediatricians, neonatologists, obstetricians, and the research community. Signature is CAP accredited, CLIA certified, and has clinical licenses from California, New York, Rhode Island and Florida. Additional information about Signature Genomic Laboratories is available at [www.signaturegenomics.com](http://www.signaturegenomics.com).

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