



A High Throughput Newborn Screening Assay For Angelman And Prader-Willi Syndromes

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The Invention

A UW researcher has developed a higher throughput method for detection of the methylation pattern of the alleles involved in Prader-Willi and Angelman disorders from dried blood spots. This method provides a means of screening for these two genetic disorders in newborn dried blood spots at state testing labs. By detecting the existence of the methylation patterns indicative of the syndromes, treatment can begin early in the lives of these children. Mei adapted/optimized a standard PCR-based method for measuring the methylation patterns at this location on the chromosome. She developed primers for the methylated and unmethylated alleles, adjusted the denaturing temperature on the thermocycler used during the PCR process, and eliminated uracil-DNA glycosylate and deoxyuridine triphosphate (dUTP) from the PCR reaction mixture.

Tech Fields

- [Diagnostics & Biomarkers : Diagnostics](#)

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