

Title: Development of a metabonomics-based approach to diagnose and classify	
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Outline of Research	The study seeks to develop a molecular profiling approach to determine if metabonomic signatures may be used for the diagnosis and classification of autism. Successful development of such a tool will enable an autism diagnosis to be confirmed earlier, appropriate interventions implemented and therefore optimise outcomes for individuals with autism. This work will recruit and study a cohort of individuals with autism and their typically developing siblings as a comparator group. The specific aims of the study are: a) To study the urinary metabolic profiles of children with autism and to explore the feasibility of applying metabonomics in the diagnosis and classification of individuals with autism, b) To establish an autism phenotyping tool to identify distinct subtypes of autism, c) To link metabonomic data with clinical and observational data in an established autism phenotyping tool to enable molecular phenotyping of autistic categories, d) To identify distinctive compounds in metabolite profiles that are characteristic of autism and/or unique to the subtypes previously identified, e) To relate distinctive compounds in the metabolite profile to the pathophysiology of autism.
Findings to date	Establishment of an autism phenotyping tool has commenced using data clinical and observational from 120 children. Recruitment for participants in the metabonomic arm of the autism study commenced in August 2005 and 60 children with autism and 60 typically siblings have had 2 morning urine samples collected. Pilot studies have been conducted to optimise analytical conditions and to determine stability of samples during storage. Samples have been processed on an API3000 LC/MS/MS. The resulting spectra, 50 m/z to 400 m/z have subsequently been subjected to pattern recognition (PR) analyses using both unsupervised and supervised PR approaches in order to characterise samples based on the properties of the spectral data. Preliminary results have been promising.
Expected start and finish dates	April 2005 to December 2010