

EXECUTIVE SUMMARY [NON-TECHNICAL ABSTRACT FOR PUBLIC INFORMATION OR PROGRAM PROMOTION]:

State **in layman's terms** the application's broad, long-term objectives and specific aims, making reference to the potential public benefits of the project relevant to California.

Chromosome abnormalities are a significant source of clinical problems observed in patients, including those with mental retardation and autism. Duplications and deletions of different segments of human chromosomes have been catalogued in association with various syndromes, some of which have been well characterized and others not. The purpose of this study is to investigate in greater detail the changes in the copy number of genes in the chromosome region of the short arm of chromosome 8, referred to as 8p, that appear to be present in association with specific clinical symptoms. Thorough cognitive and behavioral assessments will be conducted on each patient in the study, and the genetic analysis of these same individuals will be conducted by using a new DNA array technology that allows refined investigation into details of changes in gene copy number that is not possible through the conventional method of high resolution chromosome analysis. Results from this study will assist in providing a more refined diagnosis and prognosis of patients that are categorized as having the chromosome 8p aberration, and may help to home in on the genes that are responsible for specific clinical features associated with the disorder. A more detailed understanding of the underlying genetic cause of the symptoms will hopefully lead to better therapeutic methods to manage and treat the condition.