early developments Frank Porter Graham Child Development Institute

Summer 2004 | Volume 8 #2

Fragile X Syndrome


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news

FPG project featured on CNN show

The Abecedarian Study, a long-running FPG research project, was highlighted in a recent prime-time CNN show, "The Gap—50 Years After the Brown Decision." Dr. Frances Campbell, who directs the study, was featured in the show.

The Abecedarian Project is a carefully controlled study in which 57 infants from low-income families were randomly assigned to receive early intervention in a high quality child care setting and 54 were in a non-treated control group. The treated children received full-time educational intervention in a high quality child care setting from infancy through age 5.

Investigators have now completed a young adult follow-up assessment of study participants. At age 21, cognitive functioning, academic skills, educational attainment, employment, parenthood, and social adjustment were measured. Major findings of the young adult follow-up include these:

- Young adults who received early educational intervention had significantly higher mental test scores from toddlerhood through age 21 than did untreated controls.
- Enhanced language skills in the children appears to have mediated the effects of early intervention on cognitive performance.
- Reading achievement scores were consistently higher for individuals with early intervention.
- Mathematics achievement showed a pattern similar to that for reading, with treated individuals earning higher scores.

More information about the Abecedarian Study may be found at <www.fpg.unc.edu/~ABC/>

|ed| contents

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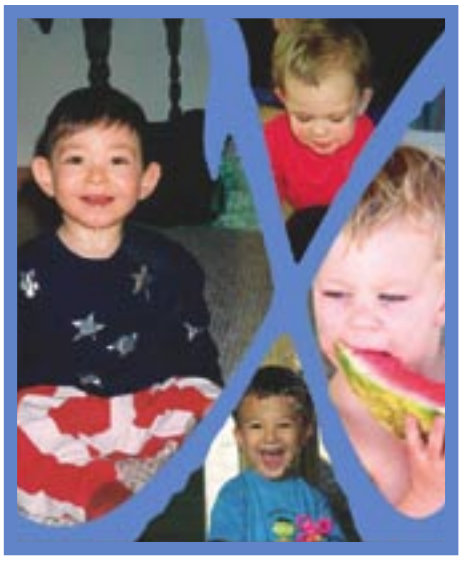
Director's Notes by Don Bailey



In 1993, some colleagues and I began a study of a little-known disorder—fragile X syndrome. The gene for fragile X had just recently been discovered, and there was lots of excitement, for many reasons. Although the disorder had been described for more than 20 years and it was almost certainly inherited, what went wrong and how it was inherited was not known. Officials at the NIH were excited because it was one of the first examples of how the Human Genome Project, a massive effort to "map" the human genome, could lead to discoveries pinpointing the causes of hundreds, if not thousands of disorders. Genetic researchers were excited because this discovery helped them to understand an unusual inheritance pattern that could not previously be explained. Many parents were excited because they finally had a better understanding of the disorder affecting their child. And the fact that fragile X is a single gene disorder gave hope to many that with the advent of gene therapy, a cure might be possible.

But of course, as with most discoveries, the answer to one question opened the door to many others. Knowing a gene flaw or "mutation" doesn't mean that you know everything about how it works. Genes, which are made up of DNA, provide instructions for making RNA, which then leads to proteins, which control all aspects of human biological function. Fragile X syndrome affects the production of a single protein (FMRP) known to be necessary for normal brain development. While much progress has been made in understanding the biology of fragile X syndrome, many mysteries remain unsolved, and a cure seems just as far away today as it did in the early 1990s.

Of course, we don't do basic biological research at FPG. But we do study children's development and behavior over time, and we are very interested in both the biological factors and the environmental factors that affect behavior and development. When we started our research in 1993, no one had studied the earliest development of children with fragile X. So we began an early childhood study, starting with children in North Carolina, South Carolina, and Virginia under the age of six. Finding nearly a hundred families of young children with fragile X was quite a challenge. But with the help of a great research team and lots of people willing to help, we did it. We have been tracking these children ever since, and some of the oldest are now getting ready to enter high school. Along the way we have involved professionals from many different disciplines and have examined the many ways in which fragile X can affect both children and families. These studies are still under way, and we are now in the midst of planning a very large study to screen 1,000,000 newborns for fragile X.

This work has been immensely gratifying. Many different people at FPG and across the university are now actively involved in fragile X research. With a strong publication record and the most comprehensive longitudinal study of fragile X ever conducted, we are well positioned to continue this work into the future. However, what we have learned from all of this goes well beyond our publications and well beyond fragile X.

- Biology is just as complicated as behavior. Compared with human development, which is enormously complicated and almost defies a "scientific" explanation, I thought biological function was relatively straightforward. In fact, genes regulate a complicated biological system with many different interactions and pathways that may never be fully understood. Although fragile X was thought to be a single gene disorder involving a single protein, in reality, it likely regulates and interacts with many other genes and many other proteins in a complex and changing pattern.
- No one person or one discipline can fully understand any phenomenon. Over the years we have worked with psychologists, neurologists, psychiatrists, special educators, genetic counselors, anthropologists, speech and language pathologists, occupational therapists, and many others who have helped to provide insights into the multifaceted aspects of this disorder.
- A focus on a single disorder still allows you to address "big" issues. For example, we now have the technology and knowledge to identify many disorders earlier than we currently are. Is it worth the investment needed to identify disorders earlier? Our work on newborn screening for fragile X will serve as a prototype for early detection of other disorders.
- You don't always know where your research will lead you. We started with a small study looking at some focused areas of development. One thing that has led to our success has been our willingness to look beyond the parameters of what we were funded to do. When we find something interesting, we study it.
- Good research takes a long time. Of course, we would all like immediate answers to questions. After more than 10 years, you would think we would have fragile X figured out. Although we know a lot more now than we did when we began, much remains to be learned. This is especially true when you do research that looks at how lives change over time.

I cannot end this column without acknowledging several groups of people. First, are all of the individuals who have worked on this project throughout the years. I hesitate to mention names, because there are too many, but you know who you are. Suffice it to say that this has been a true team effort.

Second, we appreciate the support of many different agencies and foundations. We have received funding from the Office of Special Education Programs, the National Institute for Child Health and Human Development, the FRAXA Foundation, the National Fragile X Foundation, and Ronald McDonald House Charities.

Finally, throughout this project, we have had the privilege of getting to know a group of remarkable families. They have allowed us to study just about every aspect of their children's lives and now their families' lives as well. They have been amazingly tolerant and giving of their time and energy, hoping that this work will ultimately pay off. And they have inspired us to continue. We dedicate this work to them. |ed|