



*Families and  
Fragile X Syndrome*

U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES  
PUBLIC HEALTH SERVICE  
NATIONAL INSTITUTES OF HEALTH

Dear families—

Welcome to the National Institute of Child Health and Human Development (NICHD) family album about Fragile X syndrome. As a health research agency, the NICHD family includes not only scientists, researchers, and health care providers, but also children, adults, and families who benefit from health research advances.

Finding out that your child, a member of your family, or a friend has a “syndrome” can raise some tough questions. What does having that syndrome mean for the individual? What special needs will this individual have? How will those needs affect my family? Will my child, family member, or friend be able to take part in everyday activities and important events?

Through its research, the NICHD strives to find answers to these questions. This booklet is designed to give you and your family some general information about Fragile X syndrome, its causes, its features, and its treatments. This publication also describes some of the research directions currently underway to learn more about Fragile X.

The information presented here will give you and your family a foundation of knowledge that will help in understanding options, making decisions, and finding help.

As you read, you’ll notice many photographs of members of the NICHD family. These people are not models or actors. They are individuals with Fragile X syndrome and their families. Through these images, you’ll see that even though Fragile X affects families, it doesn’t keep them from doing things that all families do—laughing, sharing, encouraging, and loving.

I hope this booklet will help your family move forward in meeting the challenges of Fragile X syndrome together.

Sincerely yours,

A handwritten signature in black ink, appearing to read "Duane Alexander". The signature is written in a cursive, flowing style.

Duane Alexander, M.D.

Director, NICHD

# Table of contents

<b>What is Fragile X syndrome?</b> .....	<b>1</b>
<b>What causes Fragile X syndrome?</b> .....	<b>2</b>
What keeps the <i>FMR1</i> gene from producing FMRP in Fragile X syndrome? .....	4
<b>Human cells 101</b> .....	<b>8</b>
<b>How many people are affected by Fragile X syndrome?</b> .....	<b>10</b>
<b>How is Fragile X syndrome inherited?</b> .....	<b>12</b>
<b>What are the signs and symptoms of Fragile X syndrome?</b> .....	<b>14</b>
Intelligence and learning .....	15
Physical .....	17
Social and emotional .....	19
Speech and language .....	20
Sensory .....	22
<b>Is there a cure for Fragile X syndrome?</b> .....	<b>24</b>
<b>Are there treatments for Fragile X syndrome?</b> .....	<b>24</b>
Educational options .....	24
Therapeutic options .....	27
Medication options .....	30
<b>What are the options for adults who have Fragile X syndrome?</b> .....	<b>32</b>
<b>What should I do if I find out someone in my family has Fragile X syndrome?</b> .....	<b>34</b>
<b>What is being done to develop treatments or a cure for Fragile X syndrome?</b> .....	<b>38</b>
<b>Where can I go for more information about Fragile X syndrome?.</b> .....	<b>42</b>
The National Institute of Child Health and Human Development (NICHD) .....	42
National organizations .....	43
Additional resources .....	44
<b>References</b> .....	<b>45</b>