

HOW TO CONQUER A CHROMOSOME ABNORMALITY

“The best thing you can do is to place that baby in an institution and then go on with your lives.”

—Quotes such as this were at one time commonly expressed by doctors to parents.



Dylan [18q-]

ONE OF EVERY 180 BABIES IS BORN WITH A chromosome abnormality, and we don't know how to prevent it. It could happen to any child. The syndromes of chromosome 18, like Down syndrome and other chromosome abnormalities, were thought to be incurable and untreatable. Today medical science has the tools to understand and treat these syndromes, but the required research needs to be done. Research is costly, so we are asking for your help.

The Chromosome 18 Registry & Research Society is a non-profit, tax-exempt public charity. We depend on contributions from individuals, businesses and philanthropic foundations to support our work. Our mission is to help individuals with chromosome 18 abnormalities overcome the challenges they face so they might lead happy, healthy and productive lives.

Your contribution can make the difference.

*Help us conquer the chromosome 18 syndromes.  
Please give generously to support research.*

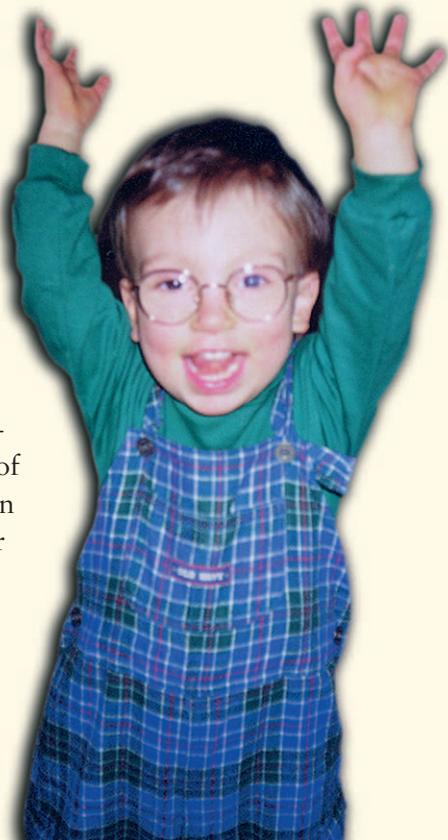


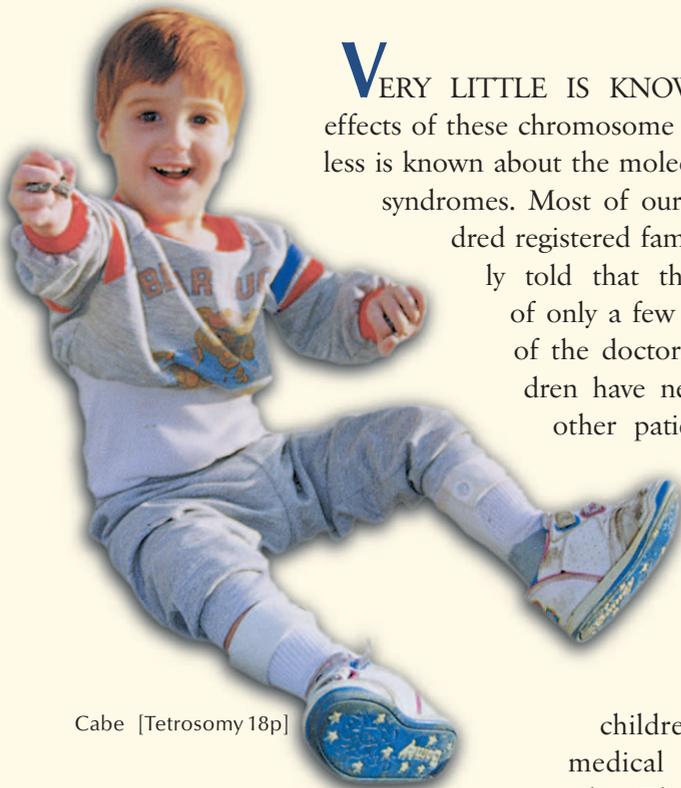
Kirsten [Trisomy18]

**O**UR CHILDREN EACH HAVE A syndrome that we are told is impossible to treat. We are told that all the chromosome 18 syndromes are too complicated to fully understand and too rare to adequately study. Our children have learning difficulties compounded by hearing impairments. They have speech delay compounded by poor muscle tone. They have multiple handicaps. But they also have determined parents! We have united to form **The Chromosome 18 Registry & Research Society**, a nonprofit support group of families whose

children have abnormalities of chromosome 18. Our purpose is to find affected families, fund research into these disorders, and get the newly gained knowledge back to the families.

**N**EW TECHNIQUES IN MOLECULAR biology, which allow identification and isolation of specific genes, now make possible the understanding of complicated conditions such as chromosome abnormalities and mental retardation. We *will* understand the syndromes of chromosome 18. We *will* see the day when treatments are available. We *will* help our children. To do this, we need your help.



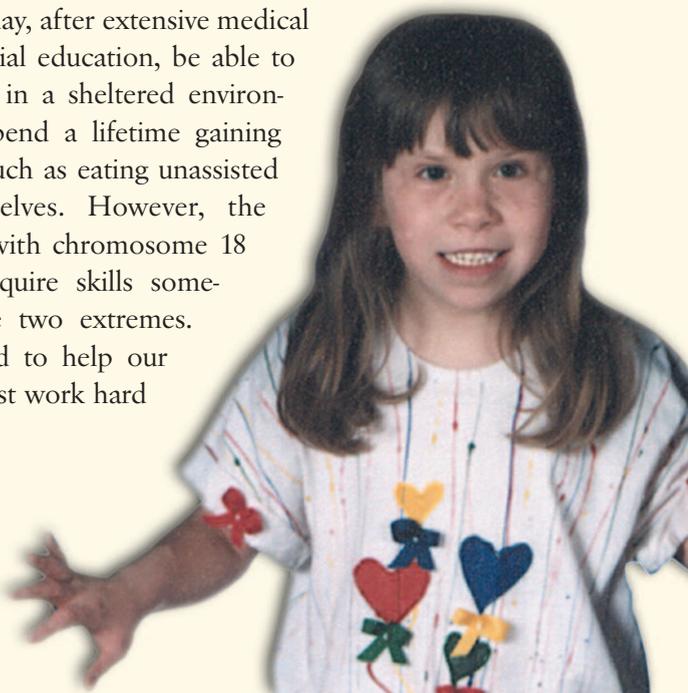


Cabe [Tetrosomy 18p]

**V**ERY LITTLE IS KNOWN ABOUT THE effects of these chromosome abnormalities. Even less is known about the molecular basis for these syndromes. Most of our nearly fifteen hundred registered families were mistakenly told that their child was one of only a few in the world. Most of the doctors who see our children have never seen even one other patient with the same syndrome. Our families need to see the experts.

his or her syndrome. The needs of children with the same syndrome can also vary greatly. All of the children have speech delay. Some will one day, after extensive medical intervention and special education, be able to live on their own or in a sheltered environment. Others may spend a lifetime gaining basic self-help skills such as eating unassisted and dressing themselves. However, the majority of children with chromosome 18 abnormalities can acquire skills somewhere between these two extremes. The answers we need to help our children exist! We must work hard to find them.

Nikki [Ring 18]



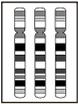
**A**LL OF OUR children have complex medical and educational needs. The extent of each child's disability is dependent upon

## WHAT IS A CHROMOSOME ABNORMALITY?

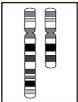
A chromosome abnormality is the loss, duplication, or rearrangement of chromosome material that is large enough to be seen with a microscope. Chromosome abnormalities actually are a common occurrence. However, only a very few allow the fetus to develop sufficiently to result in a live birth. As many as fifty percent of conceptions have chromosome abnormalities, but ninety percent of these embryos are lost very early and do not even become recognized pregnancies. More than half of first trimester miscarriages are caused by chromosome abnormalities of the fetus. The developing human body can tolerate the loss or duplication of only certain small parts of certain chromosomes and still be born. There are no known risk factors which put someone at an increased chance of having a child with a chromosome abnormality. It could happen to anyone, and it has happened to our children.

## WHAT ARE THE SYNDROMES OF CHROMOSOME 18?

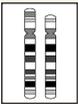
*The most frequent abnormalities of chromosome 18 are these:*



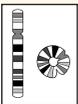
**Trisomy 18** There are three copies instead of the usual two.



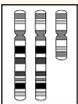
**18q-** There is a missing piece from the long arm of chromosome 18.



**18p-** There is a missing piece from the short arm of chromosome 18.



**Ring 18** One of the copies of chromosome 18 forms a ring, and material is lost from both the long and short arms.

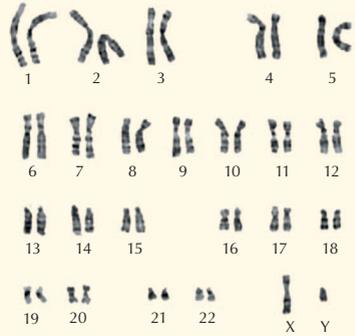
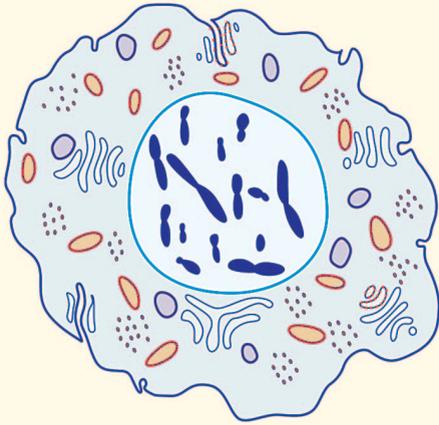


**Tetrasomy 18p** An extra chromosome is present. This chromosome is made up of two copies of the short arm of chromosome 18.

*There are many individuals with other unique rearrangements of chromosome 18.*

**CELL**

**NUCLEUS**



Every cell has a nucleus which contains chromosomes. There are 23 pairs of chromosomes in the nucleus of each cell.

Every organ and tissue in the human body is made up of cells.

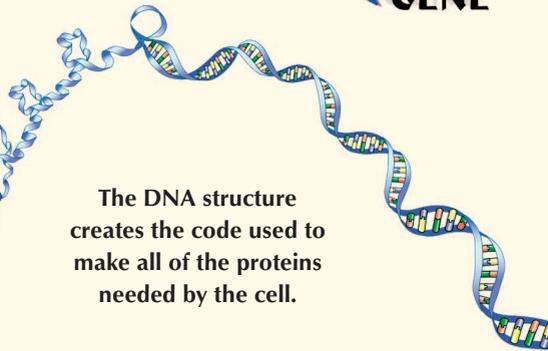
**CHROMOSOME**

**DNA**

**GENE**



Each chromosome is made of a very long strand of a chemical known as DNA.



The DNA structure creates the code used to make all of the proteins needed by the cell.



## THE CHROMOSOME 18 CLINICAL RESEARCH CENTER

is located at The University of Texas Health Science Center at San Antonio. The Center seeks to continue to:

- ◆ Provide definitive medical and educational resources for the families of individuals with chromosome 18 abnormalities.
- ◆ Perform and facilitate groundbreaking clinical and basic research relating to the abnormalities of chromosome 18.
- ◆ Provide treatments to help these individuals overcome the effects of their chromosome abnormality.

The Center involves investigators from diverse medical subspecialties at the Health Science Center. Center investigators also collaborate with leading clinicians and experts worldwide to better serve the chromosome 18 community.

**The Chromosome 18 Registry & Research Society** supports the Chromosome 18 Clinical Research Center enrollment process. This includes the medical records and DNA banks. Individuals who are enrolled will have the option of participating in studies such as surveys, medical evaluations or treatment trials as such studies are developed and funded.

*Your tax deductible contribution  
can make a difference!*

**YOUR SUPPORT BRINGS  
THE DREAM OF TREATMENTS  
FOR THE ABNORMALITIES  
OF CHROMOSOME 18  
ONE STEP CLOSER  
TO REALITY.**

*You can make a contribution  
by supporting  
one of our fund-raisers  
or by sending your donation directly to:*

**THE CHROMOSOME 18 REGISTRY & RESEARCH SOCIETY  
7155 OAKRIDGE DRIVE  
SAN ANTONIO, TEXAS 78229**

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