Barth Syndrome Foundation
2006 Scientific/Medical & Family Conference

Session:

JEANS TO GENES OVERVIEW: WHAT’S IN YOUR GENES?

Iris L. Gonzalez, Ph.D.
Thursday, July 6
• Each individual is unique -- we are loaded with differences that make us unique

• Differences are coded for in our DNA:

• **Many** changes in DNA do not cause disease and are usually located in space between genes, although some are within genes

• **A few** changes in DNA cause disease
  These are usually located within the genes or very close to them
• Genes do not exist or act in a vacuum: they, and their products, interact with other genes and their products -- they “network”

• Some genes have more than 1 function

• Some genes work only in some tissues, some in all tissues or organs

• Some genes only work at specific times during our development and lifetime, while some work all the time
Just like genes, people do not live in a vacuum but they interact with their environment and are affected by it

• How do various foods affect them?
• How do infectious agents affect them?
• How do weather and pollutants affect them?
• How does activity affect them?
• How do medications affect them?
When there is disease,

Is it environmentally caused?
Like an infectious disease,
or something you ate,
or an allergy?

Or is it intrinsic to the individual?

Is it genetic?
Our health is a manifestation of how well our genes are working and how well we get along with our environment

“NATURE and NURTURE”
All amino acid substitution mutations found occur at invariant & conserved positions in tafazzin protein.
50: leucine → proline

54: isoleucine → asparagine

57: arginine → leucine
Exon 8

Glycine 197

"hotspot"

GGG

Arginine

AGG

Tryptophan

TGG

Valine

GTG

Glutamic

GAG
Amino Acid Structures

Notice the many different shapes and sizes.