Hearing Loss and Connexin 26

Charles Williams, MD
Division of Genetics and Metabolism
University of Florida
Congenital deafness

Hearing loss at age 65

Congenital deafness
Syndromic (30%)

Non-syndromic autosomal dominant (12%)

Non-syndromic X-linked, mito, others (2%)

Non-syndromic autosomal recessive (56%)
Autosomal Dominant

Autosomal Recessive

X-linked

Mitochondrial
Autosomal Recessive Inheritance
25% risk for each child to inherit disorder
Carrier parents are asymptomatic
Equal sex distribution
Autosomal Dominant Inheritance

- 50% risk for each child to inherit disorder
- Equal sex distribution
- Carrier parent is usually affected with the disorder
Mitochondrial Inheritance
Only females transmit the disorder  
100% of offspring are at risk to show the disorder  
Variable expression and time of onset
Complex Inheritance
Multifactorial Inheritance

- Parents and siblings may not be affected
- Low sibling recurrence risk (3-10%)
Mother and father have deafness
Mother and father have deafness
Mother and father have deafness
**Autosomal Recessive**
25% risk

**X-linked Recessive**
25% risk

**Autosomal Dominant**
Spontaneous mutation
0% increased risk

**Mitochondrial**
asymptomatic mother
as high as 100% risk

**Autosomal Dominant**
asymptomatic mother
as high as 50% risk