

USHER SYNDROME



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USHER SYNDROME

- *Type I*
- *Type II*
- *Type III*

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USHER SYNDROME

- Genetically Inherited
- Sensorineural Hearing Loss
- Progressive Vision Loss - RP
- Balance Problems – types I, III

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USHER SYNDROME

- 3 – 10% Childhood deafness
- 10 – 20% RP cases

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CRITICAL FACTORS

- Early Diagnosis
- Education
- Career Guidance
- Acquisition of New Skills
- Individual and Family Counseling

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COMPARISON HEARING LOSS

Hearing Loss	I	II	III
Born	Profound Deafness	Hard of Hearing	Normal or Mild Loss
Audiogram	Corner	Sloping Sensori-neural	Sloping Sensori-neural
Response	Very loud Low tones	Mild-low freq S/P- high freq	Progressive

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COMPARISON HEARING LOSS

Hearing Loss	I	II	III
Localization	Bilateral	Bilateral	Bilateral
Balance	Absent Inner Ear Balance	Normal Inner Ear Balance	Some Balance Problems – Progressive

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COMPARISON RETINITIS PIGMENTOSA

Vision Loss	I	II	III
Night Blindness	Young Child Can be as early as 4-7 Years old	Young Child Can be as early as 6-8 Years old	Older Child to Teens Can be as early as 10 years old

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COMPARISON RETINITIS PIGMENTOSA

Vision Loss	I	II	III
Blind Spots	Early Teens	Late Teens Early Adult	Early – Mid Adulthood
Reduced Visual Field	Legally Blind Mid Adulthood	Progressive Very Individual	Progressive Very Individual

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GENETICS

- Usher syndrome is transmitted genetically by an autosomal, recessive gene. Both parents must carry the same recessive gene.

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GENETICS

- Autosomal implies that the gene is not sex linked. Therefore both males and females are equally affected.
- Recessive inheritance implies that both parents carry the same gene but are unaware of it until their child is diagnosed.

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GENETICS

- The child with Usher syndrome has two recessive genes for the trait and will transmit these genes to their children.
- If a person with Usher marries another person with the same affected gene, all the children are expected to have the same condition.

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GENETICS

- If a person with Usher marries a person without the affected gene the children may not have the condition but will be carriers.
- If a person with Usher marries another person who is deaf, the latter should be tested to determine if they carry an Usher gene.

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IDENTIFICATION

- Early identification is critical.
- If a child is identified as being in the high risk category for Usher syndrome, he/she should be referred immediately to an Ophthalmologic clinic which has the capacity for full diagnostic testing.

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