WHAT IS USHER SYNDROME?

Usher syndrome is a rare genetic condition that causes hearing loss, progressive vision loss, and possible vestibular dysfunction.

HEARING LOSS

Hearing loss occurs in people with Usher syndrome due to the hair cells in the inner ear (stereocilia) not functioning properly. As a result, the auditory nerve and the brain do not receive the proper signals and therefore unable to process sound.

VISION LOSS

The eye condition associated with Usher syndrome is *Retinitis Pigmentosa* (RP). RP is caused when the cells in the back of the eye (the retina) do not function properly, resulting in difficulties seeing in the dark and a reduced field of vision (tunnel vision).

VESTIBULAR DYSFUNCTION

Some people with Usher syndrome experience vestibular dysfunction, where the hair cells in the inner ear do not function properly. As a result, they may experience delayed gross motor development, unsteady movements, and balance issues.

HOW IS USHER SYNDROME INHERITED?

Parents of children with Usher syndrome each carry one copy of an Usher gene.

When both parents are carriers, there is a 25% chance their child will have Usher syndrome; a 50% chance their child will be a carrier like them; and a 25% chance their child will not have an Usher gene at all. This pattern of inheritance is called *autosomal recessive inheritance*.

affects 1 in 6000 individuals globally

Usher carrier frequency 1 in 70

affects 5-9% children with hearing loss

no cure

Early diagnosis enables supports to improve the quality of life for those affected

most common cause of genetic deafblindness

WHAT ARE THE DIFFERENT TYPES OF USHER SYNDROME?

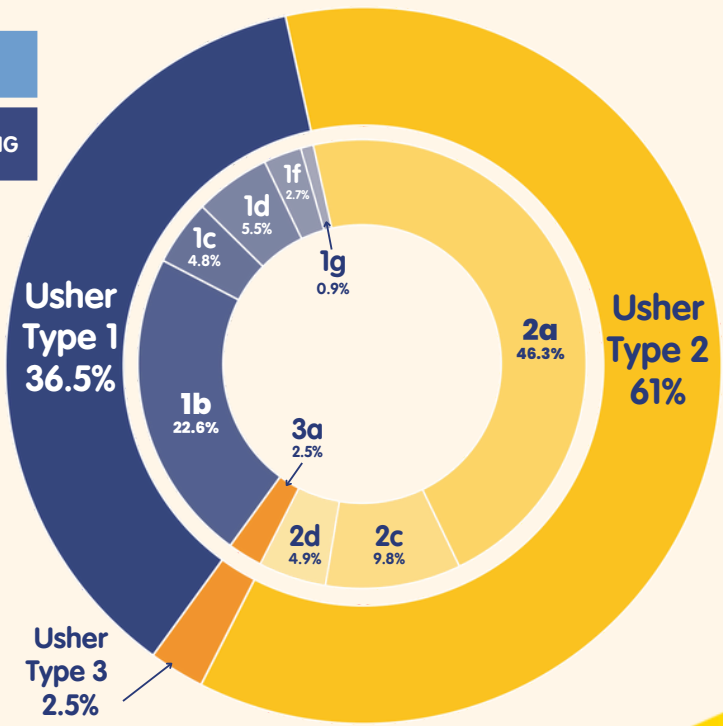
	HEARING LOSS	VISION LOSS	VESTIBULAR
TYPE 1	profound from birth	early teen onset	dysfunction from birth delayed gross motor development
TYPE 2	moderate to severe from birth	late teen to early 20's onset	normal function
TYPE 3	post-newborn onset progressive	variable	variable

USHER GENES

There are currently 9 genes known to cause different subtypes of Usher syndrome

TYPE 1	Subtype	1B	1C	1D	1F	1G
	Gene	MYO7A	USH1C	CDH23	PCDH15	USH1G
TYPE 2	Subtype	2A	2C	2D		
	Gene	USH2A	ADGRV1	WHRN		
TYPE 3	Subtype	3A				
	Gene	CLRN1				

PREVALENCE OF USHER TYPES AND GENES



*Figures are approximate
data source:
Castiglione & Möller (2022)

USHER KIDS AUSTRALIA

www.usherkidsaustralia.com