

Early Diagnosis of Usher Syndrome—Educational Opportunities and Challenges

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Educators of students with visual impairments have unprecedented opportunities to positively influence the lives of children who have been diagnosed with one of the three clinical types of Usher Syndrome. Until recently, the diagnosis was usually not made until students exited educational programs and experienced decreased visual fields as young adults. It is now possible to screen infants for Usher Syndrome. Issues related to the education of children and teens with Usher Syndrome have changed rapidly over the past decade as scientific advances have led to earlier diagnosis. Educators will want to keep informed of medical advances to appropriately plan individualized programs for students with Usher Syndrome. A new medical model of collaboration between educators and genetic scientists and neurologists is emerging. (Best, 2003).

To date, eight Usher Syndrome genes have been identified. The most cost-effective way to detect Usher Syndrome is to test babies who do not pass the newborn hearing screenings. Molecular genetic diagnosis of certain types of Usher Syndrome can be made at any age. (Kimberling, 2005).

Children with Type I Usher Syndrome usually do not begin to walk until the age of eighteen to twenty-four months. Type I Usher Syndrome is characterized by an early and profound hearing loss, retinitis pigmentosa, and vestibular problems. Usually these children use sign language or have cochlear implants.

Type II Usher Syndrome is characterized a moderate hearing loss in the lower frequencies and a severe to profound loss in the higher frequencies. There are no

vestibular problems in childhood, but the vestibular system becomes less reliable as the individual loses vision from the accompanying retinitis pigmentosa.

Type III Usher Syndrome has a progressive hearing loss and the progressive loss of visual fields and night blindness of retinitis pigmentosa. In Type III Usher Syndrome the vestibular system weakens as the person ages. Children with Type III Usher Syndrome may begin classes with students who are hard of hearing but may experience significant communication delays if their hearing and vision deteriorates. Educators must constantly reassess the communication needs of these children and remain open to classroom modifications recommended by the VI team members.

Parents looking for information on the Internet often read of the connection between delayed walking and profound deafness, first noted by Moller in 1989, and request that their child be tested for Usher Syndrome Type 1. Roughly half of the referrals to the Boys Town Hospital center are self-referred parents. (Kimberling, personal communication.) Earlier diagnosis brings many advantages but may also bring complex decisions for the parents of a baby with Usher Syndrome. Will the diagnosis influence their decision to have other children? Should they inform the school of their child's Usher Syndrome? Will a cochlear implant benefit their child's educational and social assimilation? Increasingly, parents are seeking the advice and guidance of educators with knowledge of concomitant retinitis pigmentosa and hearing loss.

Initial communication between the parents of young children with Usher Syndrome and educators of students with visual impairments most often begin with parent initiated discussion of the diagnosis. In my survey of 125 families of students with Usher Syndrome, parents in 119 of the families reported that they remembered little if anything that the physician said following the diagnosis with the progressive visual loss. The same number reported that they had never heard of Usher Syndrome and had never met anyone with Usher Syndrome. I repeated my survey in 2003 with different families, but with very similar results. Educators of students with visual impairments now have an important and early role in the formation of a family's acceptance of their child's Usher Syndrome and progressive visual loss. This is the time when parents learn not of loss, but of educational potential and possibilities for ongoing independence. Early diagnosis brings with it opportunities for earlier educational interventions that can academically benefit children who have Usher Syndrome and give hope to their families.

Early educational planning for children with Usher Syndrome should involve specialists who work with students who are deaf and educators of children who are blind and visually impaired. Both specializations have an equally important role in developing educational programs and managing the successful academic progress of learners with Usher Syndrome. The early infusion of skills and opportunities offered by orientation and mobility lessons, low vision devices, and proper lighting in the school environment,

can provide psychological reassurance and determine a defining role in the student's academic and social success. When introduced early in life, vision skills are regarded as a natural, supporting activity rather than something that will happen in the future when the child can no longer adequately function as a person who is hard of hearing or deaf. The result is reassuringly positive and helpful rather than something unknown and dreaded to be dealt with in the future.

The life of a child with Usher Syndrome is full of transitions. As with children and teens with retinitis pigmentosa, the vision may stabilize but then may diminish due to a reduction in visual fields. Each loss of vision brings with it a new period of mourning that is experienced by the child and the family. The preexisting knowledge of vision skills makes each transition easier, less frightening, and provides hope that the student can and will function independently. Students with Usher syndrome can use their knowledge of orientation, trailing, and sighted guide techniques when they encounter difficulty traveling due to night blindness. Children with Usher Syndrome almost always recall difficulty playing night games such as capture the flag, tag, or hide and seek. Often, these signs were noticed prior to their diagnosis and served as a first indication that something was different in their lives compared to their friends. Many students report that the diagnosis is a relief because they have an explanation of what is happening, and an opportunity to learn that many others in the world have similar diagnosis.

In recent years, the rapid scientific advances of genetic researchers in the field of Usher Syndrome have further refined definitions and prognosis for the three types of Usher Syndrome. Each type of Usher Syndrome has different educational issues that must be further individualized with each child's specific academic program. Students with Usher Syndrome may be identified and/or placed in mainstreamed classes or in schools or classrooms for students who are deaf or hard of hearing. The type of Usher Syndrome does not always determine the correct educational placement. There are many cases of siblings with the same clinical type of Usher Syndrome but differing educational placements based on their functional hearing, functional visual, and preferences. In one family, one daughter with Usher Syndrome lip-reads and prefers an integrated public school classroom. Her younger sister, who has the same clinical type and less restricted visual fields, finds that her needs are met only in classes of children who are deaf and communicate through sign language. One is a cheerleader and one rows with a crew. They were raised in the same home and had the same choices of educational options and training.

Sometimes, due to budget concerns, educational administrators ask the family to choose to be served by either teachers for children who are deaf or teachers of students who are visually impaired. It may be necessary for educators of students with visual

impairments to advocate for their participation and role in the planning and safety of the child with Usher Syndrome.

In my survey, I found that it is not unusual for parents of a child recently diagnosed with Usher Syndrome to resist the introduction of skill training offered by teachers of students with visual impairments. Of the 125 families, 78 initially wanted their child to only receive instruction from professionals skilled in deafness. The parents need help and information to understand that the appropriate educational interventions from the VI field will increase their child's independence, safety, and educational opportunities.

At a time when their classmates are expanding their world by learning to drive and staying out later at night, the teenager with Usher Syndrome is often experiencing an increasingly restrictive field of vision and night blindness making these activities more difficult or unsafe without appropriate professional intervention. In the United States teens with Usher Syndrome often pass driving tests and obtain their driver's license without restriction. The quickly administered vision test at the driving test site may not pick up the ramifications of night blindness or sudden darkening of the sky due to rain or snowstorms. Often potential drivers look into a small box that tests visual acuity but not field restriction. Driver's exams are given during the day when night blindness is not an issue. A driver with Usher Syndrome may not see a child in the road or hear an approaching ambulance or emergency vehicle siren. Even if a young driver leaves home on a sunny afternoon, a sudden storm may quickly darken the sky and reduce her useful vision. Parents of children with Usher Syndrome often permit their child to drive, especially in rural areas where public transportation is limited or unavailable. Teens with Usher Syndrome, like all teens, want to be like their peers and willingly accept this opportunity for increased freedom. It is recommended that a knowledgeable vision professional explain the student's functional vision to the parent and to the student in a realistic but sensitive manner offering hope through shared knowledge of travel skill instruction, orientation and mobility, and travel enhancing assistive devices. Public transportation should be offered as a viable alternative whenever appropriate and available.

Often ophthalmologists including retinal specialists are not knowledgeable about educational issues including travel skills, low vision devices, classroom modifications, and eventual employment potentials of people who have Usher Syndrome. In my interviews with 125 families of children with Usher Syndrome, I found that 46 families reported that the diagnosing ophthalmologist told their child to learn Braille without inquiring about the child's reading level or ability. Twenty-nine were told by ophthalmologists at the time of diagnosis that their children could not work in gainful employment as adults. Clearly, the message is for educators in the field of visual impairments to start with the basics when working with a student with any type of Usher

Syndrome. Emphasis on the possibilities available through the acquisition of travel skills, low vision training, career planning, and communication training provides hope.

The knowledge of travel skills should be accompanied by the opportunity for the student and the family to acquire the methodology, skills, and practice necessary for safe and independent travel in indoor and outdoor environments including the home. Ideally, the indoor instruction of the school campus should be offered each year prior to the start of the academic year when the campus is not session. If the student needs to learn how to travel to new classrooms during class changes it is important to introduce the new routes when the hallways are not crowded and when they are free of students. Open locker doors, books sitting on the floors, classroom doors opening into the hallway, and other obstacles must be dealt with during the school year. However, the special communication requirements of sign language for students with Usher Syndrome I or Usher Syndrome III, or quiet hallways permitting optimal use of residual hearing for students with Usher Syndrome Type II or Usher Type III, cannot occur successfully during chaotic, crowded class changes. It is recommended that the initial instruction be conducted in a quiet, not crowded school building for optimum communication and understanding to take place. After the essential communication is conveyed, the student can be introduced to increasingly congested hallways or school common areas. Orientation and mobility instructors who cannot sign fluently should use a sign language interpreter for all travel skill lessons for students who have Type I Usher Syndrome if they communicate through sign language. All travel lessons must incorporate considerations for the loss of auditory information from approaching cars, audible traffic signals, and nearby pedestrians.

Students who are regularly introduced to new travel patterns and skills learn to see orientation and mobility skills as a positive and life-enhancing tool for ongoing and increased independence and socialization. In my survey of 40 adults with Usher Syndrome who were not acquainted with orientation and mobility and travel skills during school years, 35 report that they view these skills as a dreaded and future necessity only if a visual loss makes independent travel impossible. Adults who have come to regard travel skills as an ongoing learning experience are able to transfer this practice to social and employment settings.

The parents of students with Type I Usher Syndrome are increasingly choosing cochlear implants for their children. This development, as well as many other issues, requires ongoing teamwork of educators from the field of visual impairments and those from deaf education to share their expertise and knowledge of functional limitations and options. One young man with Usher Syndrome was on a mobility lesson with an instructor who thought that his cochlear implant meant normal hearing. The student could not hear his instructor's verbal directions and was injured and terrified when he fell into a fifteen-foot

canal. But with ongoing team building and professional consultations, students can benefit from the combined knowledge of the VI field and the field of deafness.

With ongoing opportunities to benefit from the knowledge, skills and intervention provided by educators of students with visual impairments, students with Usher Syndrome are able to realize their strengths and move forward confidently with their lives and their dreams.

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