What is aniridia syndrome?

Aniridia syndrome is a congenital disease characterized by severe eye problems and vision deficits as well as a spectrum of other medical issues. Aniridia is usually detected at birth with the most noticeable feature being that the baby has dark eyes with no real iris color. Although, "aniridia" means lack of iris, this is not the most important issue with aniridia. When named many years ago, what physicians primarily noted was that the child had no iris, low vision and possible light sensitivity. Of primary importance is recognition that this is a life-long condition requiring constant attention to maintain even limited vision and address persistent accompanying health problems. For this reason it is important to know about the conditions which comprise the broader "aniridia syndrome."

In most cases, these eye and medical issues that make up aniridia syndrome can develop anytime from birth into young adulthood. Many of the issues that define aniridia syndrome are common in the general population affecting the eye (glaucoma, cataracts, corneal disease, retinal problems, low vision or blindness), brain function (e.g. autism spectrum disorders) and metabolic issues (e.g. diabetes and obesity). However, unlike a person in the general population, the child or adult with "aniridia syndrome" lives with many of these issues throughout their life, usually occurring far earlier than those in the general population, as in the case of cataracts, glaucoma and corneal scarring.

The genetic mutation produces a failure in development of the ocular globe during pregnancy and because of the underdevelopment of eye structures such as the optic nerve and retina the infant is born with vision in the ranges of low vision to legal blindness. The iris is just a thick collar of tissue that is never fully developed. On average, most have vision at the level of legal blindness (20/200) or worse; however, some do have "low vision" (20/40 to under 20/200). Also, the muscles which open and close the pupil are missing entirely. This syndrome is then passed down into each successive generation as a dominant gene trait. Every person with aniridia syndrome will then have a 50 percent chance of producing a child with aniridia syndrome with each pregnancy. For this reason, there is a need to help people who are affected visually, help research in this area, and stop the progression in family lines.

The approximate occurrence is 1/40,000 births and though relatively uncommon, because most of the conditions that accompany this syndrome are also common in the general population, furthering knowledge and research advancements will help many more people than just those with aniridia syndrome.

Aniridia results from a genetic error on the short arm of the chromosome 11p13 involving the PAX6 gene. PAX6 is known to be responsible for the development of the eye in humans and other animals; however, now we also know that it plays a role in many parts of the body including development of the pancreas and elsewhere in the digestive system, and within the central nervous system, resulting in the broad effects seen in the aniridia syndrome.

Medical conditions such as glucose intolerance, diabetes, auditory processing issues, autism spectrum disorders, metabolic issues and obesity have been associated with the aniridia syndrome yet these areas remain poorly studied and need further examination to understand how they arise and how best to treat the resulting serious medical problems arising from these aspects of the syndrome. Research projects in a number of these areas is either underway or planned at University of Virginia.