Scientists spot gene for rare disorder causing deafness, blindness

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(HealthDay)—Researchers say they have found the genetic cause of a

rare disorder that causes children to be born with deafness, blindness, albinism and fragile bones.

The syndrome is called COMMAD. It occurs when children inherit two mutations—one from each parent—of a gene called MITF. Each parent is also deaf due to another rare genetic disorder called Waardenburg syndrome 2A.

Further research is needed to learn more about the role of MITF during early development and how mutations in this gene result in the development of Waardenburg 2A and COMMAD, said researchers from the U.S. National Eye Institute (NEI).

COMMAD stands for the names of a number of conditions that affect people with this disorder. It includes missing tissue around the eye; abnormally dense bones prone to fracture; small or abnormally formed eyes; an abnormally large head; albinism (lack of melanin in the skin, eyes and hair), and deafness.

Identifying the genetic cause of COMMAD is important because deaf people commonly choose to marry other deaf persons. People who are deaf may not know that their deafness is associated with Waardenburg 2A, the researchers explained.

Deaf couples may want to consider genetic counseling prior to conceiving a child. If both potential parents have Waardenburg 2A, they risk passing mutated versions of MITF to their children, who would then have COMMAD, study lead author Dr. Brian Brooks said in a NEI news release.

Brooks is chief of the NEI's Pediatric, Developmental, and Genetic Ophthalmology section.

The study describes two unrelated cases of children born with COMMAD who inherited the two mutations of MITF from their parents.

Most people who are born deaf don't have Waardenburg 2A. Along with hearing loss, people with the syndrome have premature graying of the hair, blue eyes, fair skin and sometimes vision problems, the researchers said.

The study was published recently in the *American Journal of Human Genetics*.

More information: The U.S. National Human Genome Research Institute has more on genetic disorders.