



Acute Anterior Uveitis (AAU) Study

“Researchers uncover genetic region involved with a cause of potential loss of vision in people with AS”

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A locus on chromosome 9p predisposes to a specific disease manifestation, acute anterior uveitis (AAU) in ankylosing spondylitis a genetically complex multisystem, inflammatory disease.

Ankylosing spondylitis (AS) is a rheumatic disease of largely genetic origin, which means that it runs in families. It affects not only the joints and bones; other parts of the body can also be impacted. These include the heart, the lungs and the eyes.

In this study, Dr. Martin and her colleagues sought to uncover whether a specific gene or multiple genes are involved in susceptibility to a serious inflammation of the eye called iritis or acute anterior uveitis (AAU), in people with AS. AAU may occur in the absence of other inflammatory disease or in the presence of AS and related diseases. Since 40% of patients with AS also have AAU, the study enrolled a large proportion of participants with AS as well as AAU. AAU is potentially a serious disorder that can lead to blindness if not properly identified and treated.

Conclusion of the study

The study was able to identify the 9p chromosomal region as being implicated in AAU. In addition, several chromosomal regions associated with AS also were linked with AAU. Of note, the researchers indicate that the 9p region identified here is relatively large and contains at least 100 genes. Fine mapping studies (refined) are under way to further narrow the boundaries of this region on the chromosome.

The future

The results of the study are highly encouraging and suggest that some genes may influence particular complications of AS, such as eye involvement. However, the authors note that this relatively small genetic study will require follow-up with much larger cohorts (numbers of study participants) to confirm its findings.

This is important work on behalf of Dr. Martin and her colleagues. It represents a potentially important contribution to better understanding of the causes of AAU, which, as stated previously, is a very common problem in AS and which can lead to blindness.



AS has a strong familial component and is associated with HLA-B27—particularly in people with European heritage. In regard to AAU, it is generally estimated that among people of Caucasian origin, up to 50% of those with AAU are positive for the HLA-B27 gene. Interestingly, even though candidate genes—those located in a chromosomal region suspected of being involved in susceptibility—have been identified for AS, none as yet, in any of these genomic regions has been specifically implicated in extra-skeletal (parts of the body unassociated with the bones) manifestations of the disease.

The study employed a genome-wide scan (a “look” at all the chromosomes) to identify regions with linkage. This in itself was unique in that it was the first study to identify a genetic region for AAU using this method.

Thank you to all of those who participated in the study in response to the SAA’s request. Without your help, these important studies would not be possible.

To find out how to participate, please see page 15.