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HXR. Hydroxychloroquine Retinopathy

NIHR BioResource – Rare Diseases study project

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V1 26/03/2020

Summary

Hydroxychloroquine is a treatment for a number of autoimmune conditions (conditions in which the immune system starts to damage the body's own tissues). In some conditions, it has been shown that patients who take this drug are more likely to live for longer than those who are not taking this drug. It is fairly safe and not very expensive making it in many ways an ideal medication. However, a minority of patients taking this drug can develop damage to the retina (the nerve layer at the back of the eye which detects light allowing us to see). This retinal damage can lead to blindness that can get worse even when the drug is stopped. This side effect might occur to some extent in over 1 in 20 people who have been taking this medication for more than 5 years. Therefore, it is now recommended that all such patients are seen in specialist eye clinics every year so that the earliest signs of any damage can be detected and the medication could be stopped. This is a large burden on patients and the NHS as many thousands of patients have to be seen in eye clinics annually, even though most won't develop any problems.

The fact that some patients can take this drug for over 20 years and never develop any eye problems, and others can develop eye problems after just 5 years suggests that some people have genetic factors that predispose them to develop the retinal damage in response to the drug. This cohort aims to recruit those with retinopathy due to hydroxychloroquine as well as those who appear to be refractory to the retinopathy despite prolonged use of the drug. This will allow investigators to determine the genetic and other risk factors. If successful, we might be able to then predict, with a simple genetic test, which patients can safely go on the drug, and which patients should avoid the drug. This would reduce the number of patients who need to have regular retinal examinations and would also reduce the number of patients who develop blindness from the drug as those patients would not be given the drug to start with. Also, by identifying particular genes that might be involved, we might learn more about the

mechanisms by which retinal damage occurs, which might well be relevant to a number of other rare causes of retinal damage (including inherited retinal diseases).

Recruitment Criteria

Inclusion

Duration of hydroxychloroquine treatment greater than 5 years; age 18 years or over; ability to acquire retinal imaging data; clear presence (cases) or absence (controls) of hydroxychloroquine retinopathy

Exclusion

Duration of treatment under 5 years; age <18 years; inability to acquire retinal imaging data; equivocal findings on retinal imaging; other macular pathology (including macular dystrophy or advanced macular degeneration).