Genetics of Lowe Syndrome

To read some introductory information about genetics, visit our key topics section and select 'genetics' from the drop down menu.

Lowe Syndrome is a rare genetic condition caused by a mutation or an alternation to a single gene on the X chromosome.

As the disorder is X-linked (meaning that the condition is generally passed from a carrier mother to an affected son), Lowe Syndrome mainly affects males. As females have two X chromosomes, it is extremely rare for girls to have Lowe syndrome because both copies of the X chromosome would need to be affected. The affected gene is called OCRL, which has a role in enzyme production. The alternations to this gene prevents an enzyme (phosphatidylinositol-4,5-biphosphate-5-phosphatase) from being produced or working properly, which results in the cataracts and problems in the kidneys and brain that are common in individuals with Lowe Syndrome. The mechanism by which this gene defect causes problems in multiple organs is not fully understood, yet.

A further description of the genetics and diagnosis of Lowe Syndrome is included on GeneReviews. You can also find **more information** on the genetics associated with Lowe syndrome in the Lowe Syndrome Trust handbook.

More information on genetic testing can be found at: http://www.nhs.uk/Conditions/Genetics/Pages/genetic-testing-and-counselling.aspx