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News Release

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Gene discovery sheds light on causes of rare type of dwarfism

A gene linked to a type of dwarfism has been identified, in a development that will help to provide better diagnoses for those families affected.

Scientists have found that errors in a particular gene can cause profound growth defects that begin before birth.

Researchers led by the Universities of Edinburgh and Birmingham decoded genetic material from 208 individuals with primordial dwarfism. They identified mutations in a gene – known as XRCC4 – in six children with the disorder.

The findings add to the number of genes that are linked to the condition.

Primordial dwarfism encompasses a group of disorders that are characterised by a failure to grow properly. It results in a smaller body size at all stages of life and limited lifespan. People who are affected also have smaller heads, which makes them distinct from those with more common types of dwarfism.

Most children are not diagnosed until they are around three but doctors are often unable to pinpoint why the condition has developed.

Having a diagnosis can help with the management of the condition and reassure parents of what they can expect.

For example, cells grown in the lab with XRCC4 mutations were more sensitive to radiation, which could mean that affected children are at greater risk of cancer. They recommend that children with XRCC4 mutations minimise their exposure to X-rays to reduce this risk.

XRCC4 plays an important role in our maturing immune system and therefore these children are also advised to have their immunological function regularly checked.

The study is published in the *American Journal of Human Genetics* and was funded by the Medical Research Council (MRC).

Dr Louise Bicknell, of the MRC Human Genetics Unit at the University of Edinburgh, said: “These findings, alongside our previous work demonstrating that mutations in a closely

related gene, LIG4, also cause primordial dwarfism, gives us greater insight into the genetic and cellular factors important in human growth."

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