PRESS RELEASE FROM THE UNIVERSITY OF DUNDEE

Dundee University scientists make further medical breakthrough.

Scientists at the University of Dundee have taken a step closer to a full understanding of eczema and related allergic diseases with the discovery that people from different ethnic backgrounds have different mutations of the gene that causes the debilitating skin disorder.

Professor Irwin McLean and his team from the College of Medicine, Dentistry and Nursing at the University of Dundee, together with Dr Alan Irvine in Dublin, have used a groundbreaking new method to examine the filaggrin gene. The team made a major breakthrough last year when they reported that defects in the filaggrin gene can cause dry skin, eczema, eczema-associated asthma and other allergies. Their continued work has now shown that within the gene there can be several faults and that eczema sufferers of different ethnic backgrounds will have different faults within the gene. Their findings will be published in Nature magazine.

They have found, so far, 15 different mutations within the gene - if you have a mutation in your gene, you have a 60 per cent chance of having eczema. If you have two mutations in your gene, you have an almost 100 per cent chance of having eczema.

Of the mutations, 5 were prevalent in the European patients examined, who were mainly from the UK and Ireland, and 9 per cent of the population were shown to carry these gene defects. There are two mutations which are the most prevalent in all European people.

There were also two mutations prevalent in the Oriental populations that were tested. Four per cent of people of Chinese descent carry this mutation, meaning it could lead to eczema in more than 50 million people in the Far East alone.

Other mutations in the gene were found in single families and so are very rare or family specific.

The filaggrin gene is one of the hardest to decipher because of its repetitive pattern, but McLean's laboratory have developed a new method which analyses the gene fully and will allow all defects to be identified.

Based on the results, it is predicted that the filaggrin gene will be found to be a major gene for these diseases in the global sense.

Eczema affects one in five children in the UK alone and is just as common in most parts of the world. In the UK and Irish populations, the Dundee and Dublin groups have shown that the filaggrin gene is involved about half of the severe, difficult-totreat cases of eczema.

The knowledge that Irwin McLean's team is building will help them to develop a full picture of the disease, enable genetic testing, and take them a step closer to discovering new more effective treatments in years to come.

Irwin McLean says, "Once we cracked this exceptionally difficult gene, we were surprised to learn how many different defects in filaggrin were waiting to be discovered, not only in European people, but other populations worldwide."

"This is the most exciting and fast-moving project we have been involved in and the lab is buzzing with excitement."

REFERENCE

The group's latest breakthrough paper was published electronically in the top genetics journal, Nature Genetics, (available at <u>http://dx.doi.org/10.1038/ng2020</u>).

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