Ectodermal Dysplasias

by

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Do you take your teeth for granted? Most of us do, and beyond the occasional trip to the dentist hardly give them a second thought. And what about your hair? It might have unruly days, but be honest - does it ever really give you cause for concern?

Now a study funded by Action Medical Research has made important progress towards understanding more about a group of rare diseases affecting the growth and development of body tissue, resulting in improved treatment for sufferers - and potentially impacting on more common conditions such as skin cancer and even on the effects of ageing.

The two-year 88,000 GBP project, led by John McGrath, Professor of Molecular Dermatology at St John's Institute of Dermatology, London, focused on searching for genes that might have 'malfunctioned' in people born with inherited abnormalities of the hair, skin, teeth and nails.

This malfunction can lead to a range of conditions - 170 in all - known as ectodermal dysplasias. With each condition, the symptoms can range from mild to severe. Some sufferers cannot sweat because of the malformation of sweat glands, some may be born with missing fingers and toes, others need dentures and dental implants from childhood due to misshapen teeth, and some have fragile skin that easily cracks and bleeds.

No known cure

It's thought that as many as seven in every 10,000 births could be affected in some way and though the first ectodermal dysplasia syndrome was identified in the 1860s, there is still no known cure.

Things start to go wrong in the womb, when the baby develops its tissues and organs. The outermost layer of the developing child is called the ectoderm and it is defects in its formation that lead to ectodermal dysplasias. The ectoderm contributes to so many parts of the body - from the fingernails to the brain - that a sufferer can be affected in many ways.

And as well as the physical discomfort caused by the conditions, patients with highly visible skin and tissue disorders also have to put up with the stares of strangers, and the psychological impact can be profound.

Important findings

Professor McGrath's study has made some important finds, unearthing new genetic faults in several disorders; and about 30 of the known syndromes have been properly classified for the first time, which means that patients will be able to receive a more accurate diagnosis and better genetic counselling about how the disease can be passed on.

The research also discovered a new gene that is abnormal in individuals with another form of ectodermal dysplasia called Kindler Syndrome. It is a very rare disease, with fewer than 100 cases ever reported, and results in thin, fragile skin that blisters easily after trauma. Skin also becomes wrinkled and easily damaged by sunlight.

"Many of our findings will contribute towards the better management and treatment of a range of conditions that can make life very difficult for those affected," said Professor McGrath.

He added, "Our discoveries have opened up a whole new field of research that has relevance not only to the a study of ectodermal dysplasias, but to wider fields in health research such as the process of wound healing, the risks of getting skin cancer and the nature of skin ageing.

"Though our remit was to focus on conditions that affect relatively few people, the spin-off research from our findings could potentially impact on millions."

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