

Now is the time for dermatologists to learn genomics

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A review of the key genes affecting pigmentation could be the first step towards incorporating genomics into clinical practice to identify patients at high-risk of skin cancer. Credit: iStockphoto

Dermatologists may need to look further than red hair and freckles when identifying patients who might be genetically predisposed to skin cancer.

A University of Queensland review of the key [genes](#) affecting [pigmentation](#) could be the first step towards incorporating genomics into clinical practice to identify high-risk patients.

Associate Professor Rick Sturm, from UQ's Dermatology Research Centre, said the review was prompted by the need to provide the latest information to clinicians.

"The whole idea of how we will deliver medicine in the future is directed towards personal or precision medicine," Dr Sturm said.

"This will require clinicians to understand their patients' genetic background, so we really needed a primer for the [dermatologists](#) on the genes they should know about."

The review gives an overview of 15 genes which affect pigmentation of the [skin](#), eyes and hair or melanoma risk.

"These include the most important genes for pigment which, when they are mutated, have a manifestation of genetic disease," Dr Sturm said.

"Dermatologists might already have some knowledge of the gene associated with [red hair](#) and freckling, which is MC1R, but there are many others."

Dr Sturm said there were now three major genes associated with naevi or moles.

"One of these genes – IRF4 – has a big effect on naevi count, and recently it's been suggested the gene can have a significant effect on the type of melanoma a patient may get," he said.

"This gene usually affects a patient's presentation in the clinic – they have lots of moles as children and not so many as adults and they get their melanoma later in life.

"In the future, clinicians will have to know the genes affecting their

patients' pigmentation and naevi presentation, and that will affect how they examine or treat them."

Dr Sturm is a molecular geneticist who has been investigating gene expression, pigmentation genetics and the process of melanoma metastasis for 25 years.

"There has been such rapid progress in our knowledge on the genetics underlying [skin cancer](#) and pigmentation, but there's been little means of keeping dermatologists updated," Dr Sturm said.

"They are aware of the implications of differences in [skin pigmentation](#) for outcomes when it comes to sun-exposure, skin cancer and disease outcomes, but they need to begin to understand the underlying genetics.

"Our review is a stepping stone to incorporating genomics into the clinic to identify the patients that need management and surveillance."

More information: Stephen A. Ainger et al. Skin Pigmentation Genetics for the Clinic, *Dermatology* (2017). [DOI: 10.1159/000468538](https://doi.org/10.1159/000468538)

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