A common genetic signature has been discovered among three cancer prone rare skin diseases

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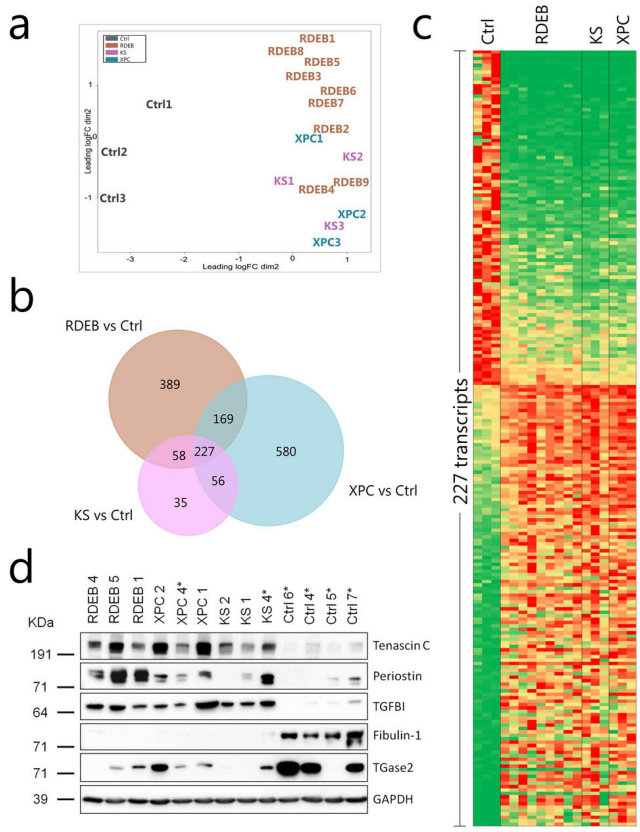

Gene expression signature in fibroblasts from RDEB, KS and XPC patients, compared to healthy donors (a). An alteration common to the three diseases of 227 genes was identified (b; c), particularly showing those associated with changes in the extracellular matrix (d). Credit: UC3M

Through a global gene expression analysis (transcriptomic), the researchers were able to find and confirm a genetic signature common to genodermatoses in patients' cells. The profile targets cellular activation and alteration of the dermal microenvironment (lower layer of the skin) which could favour the progression of the disease, as well as skin cancer.

Recessive dystrophic epidermolisis bullosa and Kindler syndrome are diseases that cause fragility of the skin, caused by mutations in essential genes to attach the two layers of the skin. Patients with these diseases suffer from chronic erosion and wounds on the skin and mucosa, which causes terrible scarring and metastatic squamous cell carcinoma to develop.

On the other hand, xeroderma pigmentosum is a disease characterised by high sensitivity to ultraviolet light, due to a deficiency in the DNA repair mechanisms, meaning patients are 110,000 times more likely to develop skin cancer.

This study, recently published in the *British Journal of Dermatology*, sheds light on the underlying molecular mechanisms of the diseases and presents new pharmacological targets that are useful for the treatment of associated effects. This possibility of treating patients therapeutically, for example, with drug repositioning, is a clinical priority in order to improve their quality of life.

More information: E. Chacón-Solano et al, Fibroblasts activation and abnormal extracellular matrix remodelling as common hallmarks in three cancer-prone genodermatoses, *British Journal of Dermatology* (2019). DOI: 10.1111/bjd.17698

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