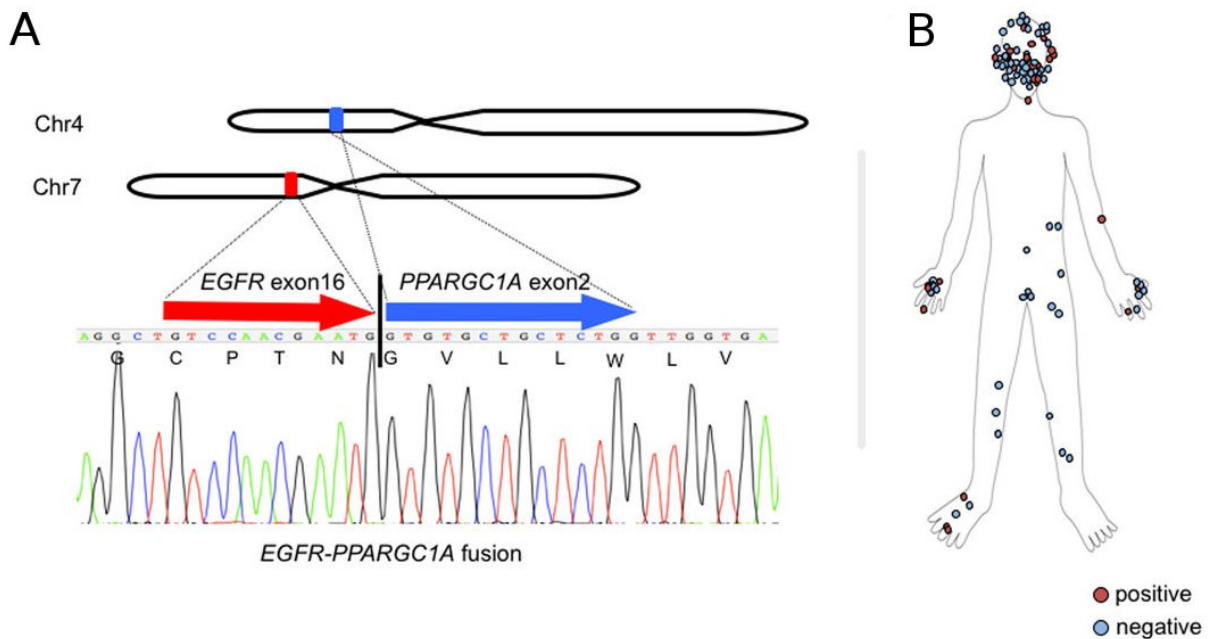


# Frequent sun exposure may cue gene fusion found in skin cancer

December 15 2017



(A) Upper: Schematic of EGFR (red) and PPARGC1A (blue) gene structures as related to EGFR-PPARGC1A fusion transcript formation. Lower: Predicted amino acid sequence and EGFR-PPARGC1A fusion transcript partial sequence at the break point. (B) The locations of cSCC tumors of 102 patients. Red dots are positive for EGFR-PPARGC1A fusion and blue dots are negative.[This image has been modified from the original (Fig. 1 & Fig. 2) of Egashira, S. et. al. (2017). Chronic sun exposure-related fusion oncogenes EGFR-PPARGC1A in cutaneous squamous cell carcinoma. *Scientific Reports*, 7(1). doi:10.1038/s41598-017-12836-z.] Credit: Dr. Masatoshi Jinnin

A fusion gene is a single composite gene resulting from the combination of two formerly independent genes. Researchers from Kumamoto University, Japan have determined that a particular fusion gene is often found in cutaneous squamous cell carcinoma (cSCC) lesions on skin exposed frequently to the sun.

Many cSCCs are easily treated by simple removal, but if left untreated they can become deadly. Current detection methods can be slow, particularly for abnormal cases, and some cSCCs are resistant to standard treatments emphasizing the need for improved diagnostics and therapies.

Fusion genes may be found in various types of connective tissue tumors, and some treatments have been developed that inhibit the tumor-forming actions of those genes. However, no [fusion](#) gene had been identified for cSCC before the Kumamoto University research team began their search. The researchers started with the cell lines A431 and DJM-1, which are frequently used in SCC studies, and normal human epidermal keratinocytes (NHEKs) as a control. Using transcriptome sequencing, they found a fusion between exon16 of epidermal growth factor receptor (EGFR) and exon2 of PPAR $\gamma$  coactivator 1- $\alpha$  (PPARGC1A) in the A431 cell line but not in the normal skin samples or other tumor tissues.

In samples of human [tumor tissues](#), the fusion gene EGFR-PPARGC1A was found in areas frequently exposed to the sun, like the face or hands, but not in areas of the body that are typically concealed in the daylight. Furthermore, [fusion genes](#) typically found in other types of SCCs were rarely found in cSCC. The researchers believe that this is because of cSCC's interrelationship with sun exposure, and that EGFR-PPARGC1A may play a part in cancers caused by ultraviolet irradiation.

"We still have more work to do to determine this fusion gene's role in cSCC tumorigenesis, but our work should provide some insights into

new drug development for this type of cancer," said project scientist, Dr. Sho Egashira. "It may be possible, for example, to assess the fusion [genes](#) in a cancerous lesion and apply a customized therapy based on the results. This would greatly reduce the risk of any side effects of the treatment."

**More information:** Sho Egashira et al, Chronic sun exposure-related fusion oncogenes EGFR-PPARGC1A in cutaneous squamous cell carcinoma, *Scientific Reports* (2017). [DOI: 10.1038/s41598-017-12836-z](https://doi.org/10.1038/s41598-017-12836-z)

Provided by Kumamoto University

Citation: Frequent sun exposure may cue gene fusion found in skin cancer (2017, December 15) retrieved 8 May 2024 from <https://medicalxpress.com/news/2017-12-frequent-sun-exposure-cue-gene.html>

<p>This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.</p>
--