

# Stabilizing Fanconi anemia with antioxidants

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Fanconi anemia (FA) is a rare genetic disorder which affects one person in 350,000. People affected by this disease have defects in DNA repair, and are hypersensitive to oxidative damage, resulting in bone marrow failure and an increased predisposition to cancer. New research published in BioMed Central's open access journal *Orphanet Journal of Rare Diseases* shows that a combination of the fatty acid  $\alpha$ -lipoic acid ( $\alpha$ -LA) and N-acetylcysteine (NAC) can stabilize the DNA of blood cells from FA patients, and drastically reduce its instability.

15 genes are known to be involved in FA. These genes are responsible for repairing DNA and because FA patients do not repair DNA efficiently they are more susceptible to diseases caused by DNA mutation, including cancer, especially leukemia. Mitochondria, the 'power houses' of the cell, are also affected by FA and consequently patients have an impaired ability to detoxify mutagenic chemicals before they can damage DNA.

Researchers from The University of Porto worked with local hospitals to obtain white blood cells from FA patients. Filipa Ponte, who performed the research, in association with Beatriz Porto and Félix Carvalho, explained, "Pre-treating cells with the antioxidants  $\alpha$ -LA and NAC (or to a lesser extent  $\alpha$ -LA or NAC individually) significantly reduced the number of spontaneous DNA breaks in blood cells taken from patients with FA and from normal controls.  $\alpha$ -LA is essential for metabolism and NAC breaks disulfide bonds and consequently is used in treating paracetamol overdose to prevent liver damage. In fact, a cocktail of  $\alpha$ -LA and NAC was able to reduce chromosome instability by at least

60%."

She continued, "These results show that  $\alpha$ -LA plus NAC cocktail may be useful to keep chromosome stability in FA patients, which could help block or delay the progression of the disease. This cocktail may be even more effective when applied to FA mosaics and FA chimeras, after bone marrow transplant, as we observed in vitro."

**More information:** Improvement of genetic stability in lymphocytes from Fanconi anemia patients through the combined effect of alpha-lipoic acid and N-acetylcysteine, Filipa Ponte, Rosa Sousa, Ana P Fernandes, Cristina Goncalves, Jose Barbot, Felix Carvalho and Beatriz Porto, *Orphanet Journal of Rare Diseases* (in press)

Provided by BioMed Central

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