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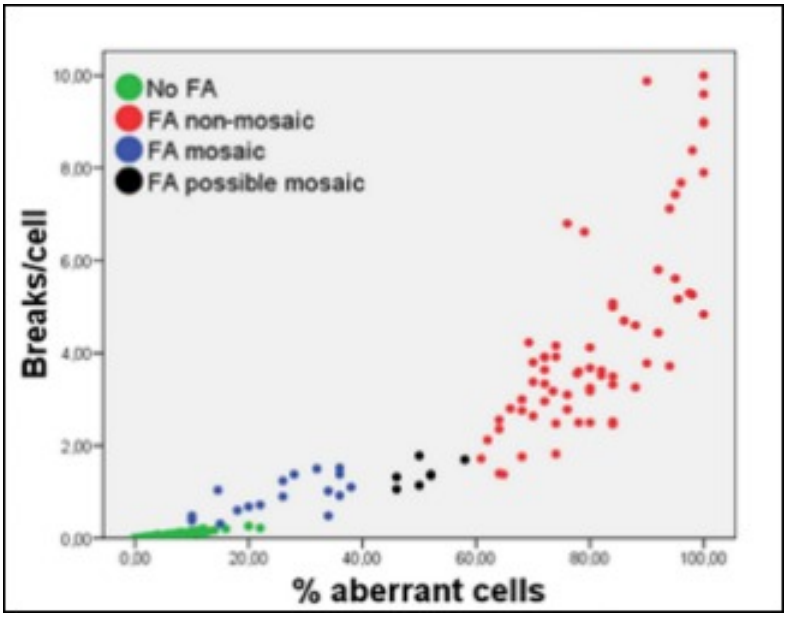
New advances in the diagnosis of Fanconi anemia



Fanconi anemia is a rare disease that causes progressive anemia and predisposition to cancer. Currently, the diagnosis based on a chromosomal fragility test is not accurate. Researchers from the UAB and the *Centro de Investigación Biomédica en Red de Enfermedades Raras* (CIBERER), in collaboration with eleven hospitals in Spain, have proposed a new index of fragility chromosome that determines a threshold level of diagnostic value to distinguish unambiguously patients with Fanconi anemia from individuals not affected.

Fanconi anaemia is a rare syndrome characterized by bone marrow failure, malformations and cancer predisposition. Chromosome fragility induced by DNA interstrand crosslink (ICL)-inducing agents such as diepoxybutane (DEB) is the 'gold standard' test for the diagnosis of Fanconi anaemia.

UAB-CIBERER researchers have investigated the variability, the diagnostic implications and the

clinical impact of chromosome fragility in Fanconi anaemia in 198 patients suspected to be affected of this genetic syndrome. This large series allowed the quantification of the variability and the level of overlap in ICL sensitivity among patients with Fanconi anaemia and the normal population.

The researchers proposed a new chromosome fragility index that provides a cut-off diagnostic level to unambiguously distinguish patients with Fanconi anaemia from non-Fanconi anaemia individuals. This study also suggests that genome instability during embryo development may be related to malformations in Fanconi anaemia, while DEB-induced chromosome breaks in T cells have no prognostic value for the haematological disease.

This study, led by Prof. Dr. Jordi Surralles (Dpt Genetics and Microbiology, Universitat Autònoma de Barcelona), been recently published in the Journal of Medical Genetics, with the participation of several research groups the pediatric hemato-oncology services of eleven Spanish hospitals (Ramón y Cajal, Madrid; Niño Jesús, Madrid; Gregorio Marañón, Madrid; Materno-Infantil Vall d'Hebron, Barcelona; Santa Creu i Sant Pau, Barcelona; Sant Joan de Deu, Esplugues; Universitario la Fe, Valencia; Reina Sofia, Cordoba; General de Las Palmas, Las Palmas de Gran Canaria; General de La Palma, Santa Cruz de La Palma; and La Princesa, Madrid).

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References

"Chromosome fragility in patients with Fanconi anaemia: diagnostic implications and clinical impact". Castella M, Pujol R, Callén E, Ramírez MJ, Casado JA, Talavera M, Ferro T, Muñoz A, Sevilla J, Madero L, Cela E, Beléndez C, de Heredia CD, Olivé T, de Toledo JS, Badell I, Estella J, Dasí Á, Rodríguez-Villa A, Gómez P, Tapia M, Molinés A, Figuera Á, Bueren JA, Surrallés J. (2011) J Med Genet. 48:242-250.

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