Fanconi Anemia and Laron Syndrome


aEscuela de Medicina, Tecnológico de Monterrey, Monterrey, Nuevo León, México
bFundación de Investigación HM Hospitales, Madrid, Spain

Background Fanconi anemia (FA) is a condition characterized by genetic instability and short stature, which is due to growth hormone (GH) deficiency in most cases. However, no apparent relationships have been identified between FA complementation group genes and GH. In this study, we thereby considered an association between FA and Laron syndrome (LS) (insulin-like growth factor 1 [IGF-1] deficiency). Methods A 21-year-old female Mexican patient with a genetic diagnosis of FA was referred to our research department for an evaluation of her short stature. Upon admission to our facility, her phenotype led to a suspicion of LS; accordingly, serum levels of IGF-1 and IGF binding protein 3 were analyzed and a GH stimulation test was performed. In addition, we used a next-generation sequencing approach for a molecular evaluation of FA disease-causing mutations and genes involved in the GH-IGF signaling pathway. Results Tests revealed low levels of IGF-1 and IGF binding protein 3 that remained within normal ranges, as well as a lack of response to GH stimulation. Sequencing confirmed a defect in the GH receptor signaling pathway. Conclusions To the best of our knowledge, this study is the first to suggest an association between FA and LS. We propose that IGF-1 administration might improve some FA complications and functions based upon IGF-1 beneficial actions observed in animal, cell and indirect clinical models: erythropoiesis modulation, immune function improvement and metabolic regulation. © 2017 The Authors

SciVal Topic Prominence
Topic: Fanconi Anemia | Fanconi Anemia Complementation Group Proteins | FA cells
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Indexed keywords

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testshort stature; signal transduction; tooth development; young adult; blood; body height; complication; Fanconi anemia; genetics; Laron syndrome; metabolism; Mexico; pathology

MeSH: Body Height; Fanconi Anemia; Female; Human Growth Hormone; Humans; Insulin-Like Growth Factor Binding Protein 3; Insulin-Like Growth Factor I; Laron Syndrome; Mexico; Receptors, Somatotropin; Signal Transduction; Young Adult

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growth hormone, 36992-73-1, 37267-05-3, 66419-50-9, 9002-72-6; protein, 67254-75-5; somatomedin C, 67763-96-6; human growth hormone, 12629-01-5;

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