

FANCONI: the word creating fuss-anemia or syndrome -a physician's dilemma

Short communication

Fanconi anemia and syndrome have been the misnomers always since these terms have been coined. The two terms seem to be very closely related but both have very varied etiologies and signs and symptoms.¹ Fanconi anemia as a term was coined way back in 1927 by the famous Swiss pediatrician Guido Fanconi. This is an autosomal recessive disorder with incidence of approximately 3 in 1,000,000.² This disease is a constellation of hereditary bone marrow failure, physical abnormalities and hyper pigmentation along with increased risk of malignancies. The basic pathophysiology lies in the increase in the chromosomal breakage, which leads to multisystem involvement.^{2,3} The marrow failure starts in the first 10 years of life and it progresses to peak upto 40-50 years of life. Approximately a third of these patients have malignancies with are not only hematological but are also non-hematological.⁴ Physical abnormalities range from hyperpigmentation, short stature to malformations of the extremities.

On the other hand, Fanconi syndrome was first described way back two decades before Fanconi anemia by Abdelhalden, though the term wasn't coined at the that time. It is basically a defect of proximal renal tubule and Guido Fanconi, on whose name we know the syndrome today as, reported 7 cases of this syndrome and called it as "Nephrotic-glycosuric dwarfism with hypophosphatemic rickets".⁵ It later came to be known as Fanconi syndrome due to a number of cases reported by Fanconi. This is a condition which is both hereditary and acquired (autosomal recessive). The basic pathophysiology in this case is the diminished absorption of the solutes by the proximal renal tubules. The patient presents with hypophosphatemic rickets, secondary growth failure, polyuria, glycosuria, acidosis etc.⁶ The complications in this case occur mainly as an after effect of renal dysfunction. So, it's the need of the hour to be clear on these two so similarly looking terms and use these with utmost care.

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Murtaza Kamal,¹ Meera Jindal²

¹Department of Pediatrics, Safdarjung Hospital & Vardhman Mahavir Medical College, India

²Department of Obstetrics & Gynaecology, Safdarjung Hospital & Vardhman Mahavir Medical College, India

Correspondence: Murtaza Kamal, Department of Pediatrics, Safdarjung Hospital & Vardhman Mahavir Medical College, C-196 (3rd Floor), Shaheen Bagh, Jamia Nagar, New Delhi-110025, India, Tel +91-9971259799, Email murtaza.vmmc@gmail.com

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Conflict of interest

The author declares no conflict of interest.

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