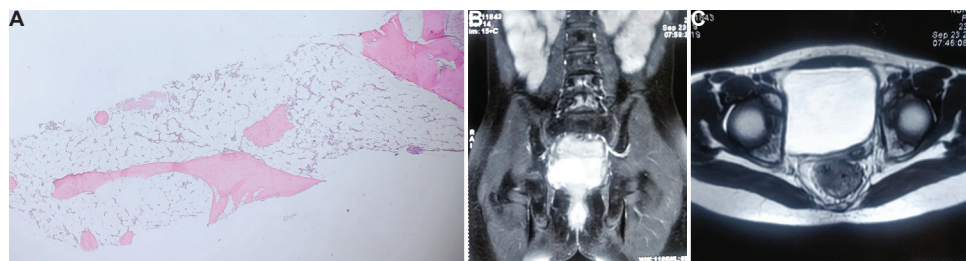




## Fanconi's anaemia & Laron syndrome: An enigma



**Figure.** (A) Photomicrograph of the bone marrow biopsy (H and E stain,  $\times 10$ ) showing hypocellular marrow with empty marrow spaces and overall cellularity of less than 5 per cent. (B) Bone marrow biopsy showing marked hypocellularity and marrow spaces replaced by fatty tissue. (C) Magnetic resonance images showing marked hypoplasia of the uterus in the transverse and cross sections.

A 12 yr old female child<sup>†</sup> presented to the Hematology outpatient department, Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, India, in September 2018 with anaemia requiring frequent blood transfusions. Haemogram showed pancytopenia. Bone marrow biopsy and aspiration were performed which revealed a hypoplastic marrow (Figure A). Chromosomal breakage analysis was negative. Next-generation sequencing for Fanconi's anaemia (FA) genetic mutations showed a heterozygous 3' splice site variation in intron 13 of *FANCA* gene.

Ultrasonogram and magnetic resonance imaging of the abdomen and pelvis (Figure B and C) revealed a hypoplastic uterus for her age measuring  $2.9 \times 0.8$  cm. In view of her short stature with delayed organ growth, tests for endocrine function were performed. The growth hormone (GH) levels were elevated while insulin-like growth factor-1 levels were low suggestive of primary GH insensitivity. A diagnosis of Fanconi's anaemia (FA) with Laron syndrome was made. She was managed with androgen therapy for the FA and was being followed by paediatric endocrinologists for further management. On one year follow up, she was

off transfusions and maintained a haemoglobin level of more than 8 g/dl.

Co-existence of these two conditions is a rare phenomena because while FA is associated with a marked increase in risk of malignancies, Laron syndrome is associated with a reduced risk. In All patients of FA, GH levels should be estimated at baseline and a thorough endocrine workup must be done.

**Acknowledgments:** Authors acknowledge Dr Muniba Alim, department of Hematology, SGPGIMS for assisting in clinical workup and diagnosis of this case.

**Conflicts of Interest:** None.

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Received November 20, 2019

<sup>†</sup>The child's assent and parent's consent obtained to publish clinical information and images.