

COMMENT ON: LUNG HYPOPLASIA AND PATELLAR AGENESIS IN EHLERS-DANLOS SYNDROME

Dear Sir,

I read with interest the article 'Lung hypoplasia and patellar agenesis in Ehlers-Danlos syndrome' by Pradhan et al.⁽¹⁾ Although the patient in question did possess numerous features of Ehlers-Danlos syndrome, I would like to suggest that the authors consider an alternative diagnosis, Meier-Gorlin syndrome.

The patient's short stature, small ears and absent patella are the classic triad of features for Meier-Gorlin syndrome.⁽²⁾ His facial appearance, such as the full lips, would also be consistent with this diagnosis.⁽²⁾ Hypermobility and joint laxity have also been described in several patients with Meier-Gorlin syndrome,^(2,3) indicating a clinical overlap with features of Ehler-Danlos syndrome. The features of the patient described by Pradhan et al⁽¹⁾ appear more in keeping with Meier-Gorlin syndrome than Ehlers-Danlos syndrome.

Pulmonary issues have been previously described in Meier-Gorlin syndrome.⁽²⁾ In addition, breast hypoplasia appears to be a consistent finding in women with Meier-Gorlin syndrome,⁽³⁾ but sexual characteristics in males have been less frequently described. Thus, this patient with lung hypoplasia, poorly developed secondary sexual characters and sparse pubic and axillary hair is an important addition to this literature. Research studies have recently discovered genes related to Meier-Gorlin syndrome,^(3,4) and genetic testing to confirm the diagnosis may be possible.

Yours sincerely,

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