Title:	New insights into the genetics of 5-oxoprolinase deficiency and further evidence that it is a benign biochemical condition
Authors:	Deshpande, A.A. (/jspui/browse?type=author&value=Deshpande%2C+A.A.) Kumar, Akhilesh (/jspui/browse?type=author&value=Kumar%2C+Akhilesh) Bachhawat, A.K. (/jspui/browse?type=author&value=Bachhawat%2C+A.K.)
Keywords:	OPLAH gene Oxoprolinase deficiency Oxoprolinuria Yeast growth assay
Issue Date:	2015
Publisher:	Springer Verlag
Citation:	European Journal of Pediatrics, 174 (3) pp. 407-411.
Abstract:	Inherited 5-oxoprolinase (OPLAH) deficiency is a rare inborn condition characterised by 5-oxoprolinuria. To date, three OPLAH mutations have been described: p.H870Pfs in a homozygous state, which results in a truncated protein, was reported in two siblings, and two heterozygous missense changes, p.S323R and p.V1089I, were independently identified in two unrelated patients. We describe the clinical context of a young girl who manifested 5-oxoprolinuria together with dusky episodes and who is compound heterozygote for two novel OPLAH variations: p.G860R and p.D1241V. To gain insight into the aetiology of the 5-oxoprolinase deficiency, we investigated the pathogenicity of all the reported missense mutations in the OPLAH gene. A yeast in vivo growth assay revealed that only p.S323R, p.G860R and p.D1241V affected the activity of the enzyme. Conclusion: Taken together, this report further suggests that hereditary 5-oxoprolinase deficiency is a benign biochemical condition caused by mutations in the OPLAH gene, which are transmitted in an autosomal recessive manner, but 5-oxoprolinuria may be a chance association in other disorders
Description:	Only IISERM authors are available in the record.
URI:	https://link.springer.com/article/10.1007/s00431-014-2397-0 (https://link.springer.com/article/10.1007/s00431-014-2397-0) http://hdl.handle.net/123456789/3121 (http://hdl.handle.net/123456789/3121)
Appears in Collections:	Research Articles (/jspui/handle/123456789/9)

Files in This Item:

File	Description	Size	Format	
Need to add pdf.odt (/jspui/bitstream/123456789/3121/1/Need%20to%20add%20pdf.odt)		8.63 kB	OpenDocument Text	View/Open (/jspui/bitstream/12345

Show full item record (/jspui/handle/123456789/3121?mode=full)

(/jspui/handle/123456789/3121/statistics)