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Correspondence

Response to “Letter to the editor”

Dear Editor,

We appreciate Dr. Turan's helpful comment and apologize for not citing the previously published abstract, which was not known to the authors.

Since conference abstracts are not visible in Pubmed they usually find their way into publications only if one of the authors attended the respective conference, which was not the case here. It is unfortunate that the clinical details of our dysosteosclerosis patients could not be compared to learn more about the fascinating mechanism of this disorder. It has become difficult to publish manuscripts on clinical details of individual cases with mutations in known disease genes.

Therefore, it is recommendable that different groups put their cases of rare disorders together to publish larger cohorts. Platforms like Genematcher have greatly facilitated the exchange of information between geneticists, but are mostly used for discovery of novel disease genes.

This case here indicates that we all should communicate more also about novel mutations in known disease genes.

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