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Response to “Germline Mosaicism in Cornelia de Lange Syndrome: Dilemmas and Risk Figures” by Mariani et al

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TO THE EDITOR

We appreciate Dr. Selicorni’s interest in our report on the contribution of germ line mosaicism to the recurrence of Cornelia de Lange Syndrome (CdLS) within families [Slavin et al., 2012]. An important point in our publication is the stated ascertainment bias in our study cohort:

This represents a highly biased sample since familial cases were sought after to give power to the initial linkage studies to identify the causative gene. Thus, this data may be skewed as the true number of familial cases in the general population is unknown.

This is important to consider, rather than simply comparing the absolute numbers for occurrence of germ line or suspected germ line mosaicism out of context.

References

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