The Role of Cilia in the Setting of Isomerism and why Isomerism is not a Subset of Heterotaxy

We read with great interest the case report by Narahari *et al*. regarding "primary ciliary dyskinesia" in a patient with left isomerism.[1] The authors do not share in this case report how the diagnosis of ciliary dyskinesia was made. They correctly share in the discussion that video microscopy to characterize ciliary beat is one such method. This is of particular importance in those with isomerism as ciliary ultrastructure can often be normal in these patients despite having ciliary dysfunction as noted by abnormal ciliary beat. Thus, electron microscopy may be normal while a ciliary function may still be impaired. This also implies that this may not be "primary ciliary dyskinesia" in its purest sense.

Ciliary dysfunction has been noted in a large proportion of patients with isomerism, with most having normal ciliary ultrastructure. Low exhaled nitric oxide levels have also been noted in those with isomerism and ciliary dysfunction.[2] While the current studies have focused on investigating motile cilia in the setting of isomerism, it is likely that sensory cilia are also dysfunctional in the setting of isomerism. Future investigations regarding this are necessary. As stated by Narahari *et al*., this abnormal ciliary function may not only be a result of isomerism but is felt to likely be implicated, at least in part, to the development of isomerism.

In addition, it should be noted that isomerism is the term that best describes this clinical situation and that isomerism is not a subset of heterotaxy. While both terms have been proposed to describe such clinical situations, the two are separate nomenclature systems. Isomerism is subdivided into right- and left-sided varieties based on the morphology of the atrial appendages while heterotaxy is subdivided by splenic anatomy. Emerging molecular data regarding this clinical situation indicates the concept of isomerism is of greater utility as the same mutation in animal models results in consistent atrial appendage findings but not consistent splenic findings.[3] We also offer that the term "situs ambiguus" be avoided. Indeed, nothing is ambiguous as the authors describe by clearly defining the location of organs which is more helpful.[4]

The nuances of isomerism are subtle but vital as we improve our understanding and management of this complex, multisystem clinical entity.

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Conflicts of interest

There are no conflicts of interest.

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