Correspondence

Establishing a relationship between clinical features and one specific type of chromosome abnormality

We read the article by Thillainathan[1] and colleagues published in this Journal. The authors provided descriptive analysis of children in Sri Lankan who underwent cytogenetic analysis for suspected chromosomal disorders. They compared their results with that in Caucasian and other Asian populations. This is an important document in that the authors added their data of Sri Lanka to the international pool of these types of surveys.[2,3] However, further analyses may benefit the impact of the current study.

The authors presented the cytogenetic outcomes in detail, but did not make an attempt to establish any possible linkage between clinical features and one specific type of chromosome abnormality. We know that signs and symptoms at presentation of some disorders may be non-specific. The clinicians and researchers try to summarize a relationship between some clinical symptoms and a specific chromosomal abnormality, just like some syndromes, i.e. a group of phenotypic symptoms in 22q11 micro-deletion syndrome. This database will benefit the clinical diagnosis greatly, and help the medical professionals to decide whether cytogenetic analysis is, or is not necessary for certain clinical features. In addition, the researchers can do more work on the genotype and phenotype for a specific type of chromosome abnormality, and also add more evidences to the database. Therefore, this is a valuable topic requiring further researches to illustrate the possible associations, and we highly recommend the authors to do deeper analysis focusing on it.

One of the limitation of the article is that the authors did not mention that if the parents of the children confirmed with chromosomal abnormalities participated into the analysis. Was the abnormality found in the child also found in the parents? For many chromosome abnormalities, parents may be carriers. It was also interesting if the parents had similar clinical manifestations to the child, with which we could know more about the relationship of genotype and phenotype for one certain abnormality, and confirm this association. We think the present analysis reflects the influence of experience of clinicians on making clinical diagnosis of suspected chromosome disorders.

In all, this is an important article in this field, but should do further analysis to benefit clinicians more.

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References

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We agree with the reader's comments that the availability of clinical features would benefit the clinical diagnosis greatly, and help the medical professionals to decide whether cytogenetic analysis is, or is not necessary for certain clinical features. However, the main aim of our study was to describe the frequency of various types of chromosomal abnormalities in Sri Lankan children undergoing cytogenetic analysis since there was paucity of data in this field. The clinical diagnosis of suspected chromosomal disorders was made by the referring clinicians and patients were referred to us for cytogenetic confirmation of the underlying chromosomal abnormalities. We therefore did not make an attempt to establish possible linkages between the clinical features and specific types of chromosomal abnormalities in all the cases, but rather documented the pattern of cytogenetic abnormalities seen in those referred for suspected chromosomal abnormalities.

Regarding karyotyping the parents of the children, we have dealt with it in the methods section, briefly it is routine practice in our lab to karyotype the parents when