Modifying Other’s Originality without Quote is an Act of Piracy

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A defective CYP21A2 gene downstream of the TNXB gene in congenital adrenal hyperplasia (CAH) falls into three categories: (a) small-scale conversions of CYP21A1P, (b) spontaneous mutations, and (c) chimeric RCCX modules that include the chimeric CYP21A1P/CYP21A2 and TNXA/TNXB genes [1]. Most of the CYP21A2 mutations identified so far were a result of small-scale conversions of the CYP21A1P (up to 11 for CYP21A1P) during both meiosis and mitosis [2], which account for about 70%-80% of all CAH cases. The Chimeric CYP21A1P/CYP21A2 and TNXA/TNXB genes, which result from unequal cross-over (or deletions) during meiosis [2] and occur in ~20% of CAH alleles in most populations [1,3] respectively reflect the deletion of the 1/XCYP21A1P - XA - RP2 - C4B - 1/XCYP21A2 gene array (1/X indicates an uncertain fraction of the gene sequence) [1] and a deletion of the RP2 - C4B - CYP21A2 - 1/XTNXB gene array [1]. Their deletion range is about a 26- or 32-kb gene sequence which depends on whether C4B is the long or short gene (more commonly shown in the literature as being 30 kb). In fact, these different types of large-gene deletions in the RCCX region are generally considered to represent one event in many studies.

I read with interest the recent article by New et al. [4], in which the authors described an analysis of 1,507 CAH families. They showed CYP21A2 gene deletion with 9 phenotypes of the chimeras, designating them CH1, CH2, CH3, CH4, CH5, CH6, CH7, CH8, and CH9 [4] in figure configuration (as attached Figure 1 in the context). However, by examining the figure, the word abbreviation of “CH” representing “Chimeric CYP21A1P/CYP21A2” was originally used by the Lee's study [1,5-8]. Furthermore, there were more modifications of the figure configuration from Lee's studies such as font including “CH-1” style (Lee’s study) (Figure 1A) having been changed into “CH1” (Figure 1B) and configuration of exons representation for the CYP21A1P and CYP21A2 genes as black boxes and white boxes respectively (in Lee's study) (Figure 1A) having been modified into CYP21A1P and

Figure 1: Diagram of the representation of the chimeric CYP21A1P/CYP21A2 genes configuration. (A) The structure of the functional CYP21A2 gene containing ten exons indicated by a white box [1,5-9]. The solid arrows indicate mutations which exist in CYP21A1P which showed the black box. CH-1 represents a chimeric CYP21A1P/CYP21A2s as described in Lee studies [1,5-9]. 707-714delGAGACTAC in CYP21A1P is marked by an asterisk (*). (B) The structure of the functional CYP21A2 gene indicated by a black box and the white box represents a nonfunctional CYP21A1P gene in New et al. study [4].

Table 1: Study on chimeric CYP21A1P/CYP21A2 gene in congenital adrenal hyperplasia.

<table>
<thead>
<tr>
<th>Study</th>
<th>Publication</th>
<th>Designation for chimeric CYP21A1P/CYP21A2</th>
<th>Ref.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lee HH*</td>
<td>+</td>
<td>CH-1 to CH-5</td>
<td>[1,5-9]</td>
</tr>
<tr>
<td>Chen et al.</td>
<td>*</td>
<td>CH-1 to CH-9</td>
<td>[5]</td>
</tr>
<tr>
<td>New et al.</td>
<td>_</td>
<td>CH1 to CH9</td>
<td>[4]</td>
</tr>
</tbody>
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*There were more than 10 articles related with chimeric CYP21A1P/CYP21A2 gene study in the past 10 years.

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References

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