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Array comparative genomic hybridisation analysis of boys with X-linked hypopituitarism identifies a 3.9 Mb duplicated critical region at Xq27 containing SOX3

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CORRESPONDENCE

Array comparative genomic hybridisation analysis of boys with X-linked hypopituitarism identifies a 3.9 Mb duplicated critical region at Xq27 containing *SOX3*

In our previous article,[1] we used array-comparative genomic hybridisation (CGH) analysis to identify Xq26–q27 duplications in families with X-linked hypopituitarism (XH). The array-CGH assay was validated using affected male genomic DNA from two previously characterised XH families that carry Xq26–q27 duplications[2 3] (fig 1B,C) and by interphase fluorescence in situ hybridisation (FISH) analysis (fig 2). Array-CGH analysis of three novel XH families, A, B and C (fig 3), indicated that each of these contained a different Xq26–q27 duplication (fig 1D–G).

Recently, we repeated these array-CGH experiments using a more extensive X chromosome array containing over 2000 bacterial artificial chromosome (BAC) clones.[4] As expected, duplications were identified in males from the two previously characterised XH families.[2 3] However, we have been unable to detect Xq26–q27 duplications in affected males from families A, B and C. Furthermore, repeated quantitative real-time PCR experiments performed by an independent collaborator in a blinded assay detected *SOX3* duplication in affected males from control families,[2 3] but not in affected males from families A, B and C. On the basis of these new data, we now cannot replicate the observation of duplications in families A, B and C families that was published in our paper.

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