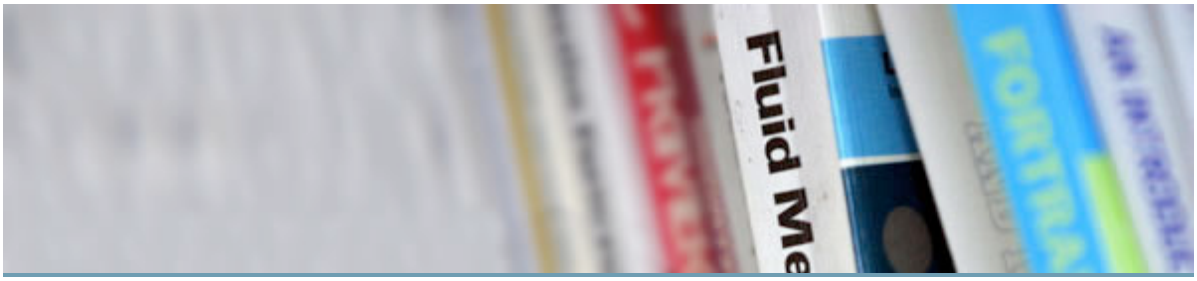

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Screening of subtle copy number changes in Aicardi syndrome patients with a high resolution X chromosome array-CGH

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Résumé / Abstract
Aicardi syndrome (AIC) is an uncommon neurodevelopmental disorder affecting almost exclusively females. Chief features include infantile spasms, corpus callosal agenesis, and chorioretinal abnormalities. AIC is a sporadic disorder and hypothesized to be caused by heterozygous mutations in an X-linked gene but up to now without any defined candidate region on the X chromosome. Array based comparative genomic hybridisation (array-CGH) has become the method of choice for the detection of microdeletions and microduplications at high resolution. In this study, for the first time, 18 AIC patients were analyzed with a full coverage X chromosomal BAC arrays at a theoretical resolution of 82 kb. Copy number changes were validated by real-time quantitation (qPCR). No disease associated aberrations were identified. For such conditions as AIC, in which there are no familial cases, additional patients should be studied in order to identify rare cases with submicroscopic abnormalities, and to pursue a positional candidate gene approach.

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