



ASSOCIATION DES CYTOGÉNÉTICIENS DE LANGUE FRANÇAISE

*Association loi 1901 déclarée au JO du 12 Août 1987
Faculté de Médecine Paris Descartes
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The ACLF fully supports the Eurogentest position paper regarding the revision of the IVD directive. More specifically, the exemption from CE-marking for in-house tests manufactured in public health service laboratories should be retained, as detailed in the Eurogentest document. However, the exemption should be restricted to laboratories accredited to ISO15189 or the equivalent. This would provide an appropriate balance of test availability and patient safety for in-house tests.

In addition to the arguments put forward by Eurogentest regarding the molecular diagnosis of rare diseases, we would like to stress the importance of in-house tests manufactured on the basis of the day-to-day practice of cytogenetics laboratories.

Indeed, it has become standard practice to investigate the structural chromosomal abnormalities revealed by karyotyping using fluorescent *in situ* hybridization (FISH) methods with specific chromosome probes. In most cases the chromosomal abnormalities are confined to a single patient or a family and cannot be detected using commercial FISH probes. This is the reason why cytogenetics laboratories are currently using in-house designed FISH probes made from publicly available BACs, fosmids or cosmids.

Furthermore, patient-specific tests are becoming increasingly common with the spread of highly sensitive whole-genome testing by array comparative genomic hybridization (CGH), for which we clearly need to check the imbalances detected with FISH in order to provide accurate genetic counselling.

Because array CGH testing reveals only genomic imbalances but not the mechanism underlying the chromosomal abnormality, we definitely need FISH testing with patient-specific BAC probes to visualize the location of the abnormality on metaphase chromosomes. When the size of the abnormality is small, cytogenetics laboratories have to design specific quantitative PCR tests to confirm the abnormality. Thus, in-house designed tests are of paramount importance for high quality testing for chromosomal abnormalities since this cannot be achieved by using commercially available tests alone.

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Should the exemption be limited to public health institutions' accredited laboratories?

We do not support this idea because this would not allow private laboratories to fulfil the requirements of the Good Practices recommendations in Cytogenetics, at least as published in France. Private laboratories should be able to design their own probes in order to complete the cytogenetic analysis provided that they are appropriately accredited.

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