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How can I confirm my (digital)MLPA results?

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We recommend confirming all abnormalities detected by conventional MLPA or digitalMLPA whenever possible, using either a different (confirmation) probemix when available or an independent technique. Copy number changes detected by a single probe should always be confirmed.

It is essential to confirm copy number changes detected by a single probe. Single probes may more readily be affected by experimental issues or variability. In addition, a mutation or polymorphism in the probe's target sequence may lead to a reduced signal, which can even mimic a deletion. Sanger sequencing of the probe's target sequence can reveal if there are any variants in the probe's target sequence in such cases.

If available, you can use a specially designed confirmation probemix that has probes with different target sequences than the primary probemix. If a confirmation probemix is available, this is usually mentioned in the product documentation. It may also be possible to confirm results obtained with digitalMLPA with a conventional MLPA probemix, or vice versa. You can also use a variety of other techniques for confirmation purposes, including Sanger sequencing of (long range) PCR products, qPCR, long-range PCR, NGS technologies, and array-CGH techniques. The method that is most suitable depends on the complexity of the region, the size of the aberration, and the availability of other techniques, among other things.

Tags

digitalMLPA

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Disclaimer

The information provided in this material is correct for the majority of our products.

However, for certain applications, the instructions for use may differ. In the event of conflicting information, the relevant instructions for use take precedence.