Reply to the Sannier et al. letter entitled “MDR3 immunostaining on frozen liver biopsy is not a sensitive diagnosis tool for the detection of heterozygous *MDR3/ABCB4* gene mutation”

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Sir or Madam,

We read with great interest the letter by Sannier et al. The data presented are consistent with our publication [1].

The authors studied a new series of adult patients with unexplained cholestasis and heterozygous *MDR3/ABCB4* mutations. In five cases, a MDR3 immunostaining was performed on liver frozen sections and showed an intense canalicular staining similar to a normal staining. These results are in contradiction with their initial report that showed a faint MDR3 immunostaining in four patients with heterozygous *MDR3/ABCB4* mutations [2]. This discrepancy may be explained by technical hazards or by the type of *MDR3/ABCB4* mutations (indeed, the mutations were different in the two series).

It could be interesting to perform MDR3 immunostainings on the formalin-fixed paraffin-embedded liver tissues

in these two series that would enable a comparison with our published data which were obtained with this technique. To conclude, it seems that *MDR3/ABCB4* heterozygous mutation diagnosis requires gene sequencing.

References

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