



Reply

Reply to Bouva et al. Comment on “Dijkstra et al. A False-Negative Newborn Screen for Tyrosinemia Type 1—Need for Re-Evaluation of Newborn Screening with Succinylacetone. *Int. J. Neonatal Screen.* 2023, 9, 66”

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We thank the authors for their comments [1]. We are happy that they agree with the content of the report [2] and underline the importance of having optimal cut-off values of succinylacetone in order to—on the one hand—not miss patients like the one presented in this report and, on the other hand, to exclude false positive results, for example, due to a MAAI deficiency. As stated in the original article (Van Vliet et al. 2023, [3]), this might mean that the succinylacetone cut-off in the newborn screening needs to be lowered, while adding additional biomarkers that can be used to distinguish children with mild Tyrosinemia type I who need treatment from those with MAAI deficiency, who do not need treatment at all.

Regarding this aim, the results from the mutual project on the improvement of newborn screening for Tyrosinemia type I, as mentioned in the comment, are needed.

We further call attention to other institutes who have children with a (suspected) MAAI deficiency to send us dried blood spots and urine to increase our knowledge on MAAI deficiency.

Conflicts of Interest: The authors declare no conflicts of interest.

References

1. Bouva, M.J.; Maase, R.E.; van Elburg, R.M. Comment on Dijkstra et al. A False-Negative Newborn Screen for Tyrosinemia Type 1—Need for Re-Evaluation of Newborn Screening with Succinylacetone. *Int. J. Neonatal Screen.* 2023, 9, 66. *Int. J. Neonatal Screen.* 2024, 10, 65. [[CrossRef](#)]
2. Dijkstra, A.M.; Evers-van Vliet, K.; Heiner-Fokkema, M.R.; Bodewes, F.A.J.A.; Bos, D.K.; Zsiros, J.; van Aerde, K.J.; Koop, K.; van Spronsen, F.J.; Lubout, C.M.A. A False-Negative Newborn Screen for Tyrosinemia Type 1—Need for Re-Evaluation of Newborn Screening with Succinylacetone. *Int. J. Neonatal Screen.* 2023, 9, 66. [[CrossRef](#)] [[PubMed](#)]
3. van Vliet, K.; Dijkstra, A.M.; Bouva, M.J.; van der Krogt, J.; Bijsterveld, K.; van der Sluijs, F.; de Sain-van der Velden, M.G.; Koop, K.; Rossi, A.; Thomas, J.A.; et al. Maleic acid is a biomarker for maleylacetoacetate isomerase deficiency; implications for newborn screening of tyrosinemia type 1. *J. Inherit. Metab. Dis.* 2023, 46, 1104–1113. [[CrossRef](#)] [[PubMed](#)]

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