Erratum: Missed Newborn Screening Case of Carnitine Palmitoyltransferase-II Deficiency

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Chapter 528: Missed Newborn Screening Case of Carnitine Palmitoyltransferase-II Deficiency, doi: [10.1007/8904_2016_528](https://doi.org/10.1007/8904_2016_528)

Andrew C. Edmondson, Jennifer Salant, Lynne A. Ierardi-Curto, and Can Ficicioglu

The print and online versions of this chapter contain some of the errors, and the corrections are given below:

Figures were reanalyzed and interpreted with the guidance of Piero Rinaldo MD, PhD. The result, tables, and discussion points also were modified based on reanalysis of figures.

Case report

Line 15: The following sentence added

Retrospective evaluation of the NBS results obtained on day-of-life 2 revealed the acylcarnitine profile also shown in Table 1.

Discussion

Line 2: Reference changed to McHugh et al. 2011

Line 3: Reference added (Gempel et al. 2002)

Line 6: Modified paragraph based on reanalysis of figures.

Our patient's (C16 + C18:1)/C2 ratio of 0.34 was indeed below the 95th percentile of the reference range for DBSs reported in that paper (0.37). We also analyzed our patient's NBS acylcarnitine profile with the productivity and post-analytical interpretive tools of the Region 4 Stork (R4S) collaborative laboratory quality improvement of newborn screening by tandem mass spectrometry (<https://www.clir-r4s.org/>) (Marquardt et al. 2012; Hall et al. 2014). This

produced the results presented in Fig. 1: (1) our patient's (C16 + C18:1)/C2 ratio was barely above the 99th percentile of the R4S cumulative reference range (0.33) but did not reach the lowest level (0.38) needed to contribute a score; (2) The overall case score was zero, but it was so because of an existing rule that forces a zero score whenever the C16 value is below the 90th percentile of the reference range ($3.44 < 4.17 \mu\text{mol/L}$). Because of this rule, which was deemed adequate based on the analysis of 110 other cases, the R4S tool would have generated a zero score both at the time of birth and 4 years later; (3) Once the rule was modified in light of this case and propionylcarnitine was upgraded to informative marker, the overall profile would generate an informative score driven by a low concentration of propionylcarnitine and four new ratios (Fig. 2). In this case, either reliance on cutoff values or the R4S tool would have failed to detect CPT-II deficiency. On the other hand, this false negative event also represents an opportunity for continuing and evolving clinical validation, as similar cases could be detected by the revised tool.

Line 51: The following sentence added

To test this hypothesis, three new ratios have already been added to the CPT-II tool in the new version of R4S named CLIR (Collaborative Laboratory Integrated Reports; <https://clir.mayo.edu>) (Fig. 2).

Table 1 modified and Table 2 deleted

Figure 1 modified and Figure 2 added

Figure 1 Legend

Partial display of the post-analytical interpretive tool for CACT/CPT-II from the Region 4 Stork (R4S)

collaborative laboratory quality improvement of newborn screening by tandem mass spectrometry website (<https://www.clir-r4s.org/>). The plot by condition panel is an overlay graph where for each marker and ratio the reference population, disease range and the case value are shown. All values are expressed as $\mu\text{mol/L}$ and converted to multiple of the normal median on a log scale. Abbreviations are as follows: *RR* reference range, *NI* not informative, *X* potentially informative score rejected by the C16 rule. Tool accessed on October 23, 2015, reproduced with permission

Figure 2 Legend

Partial display of the post-analytical interpretive tool for CACT/CPT-II from the Collaborative Laboratory Integrated Reports (CLIR) website (<https://clir.mayo.edu>). See legend of Fig. 1 for details. Additional abbreviation is as follows: *I* informative score. Tool accessed on July 15, 2016, reproduced with permission

Acknowledgements

The following sentence added:

The authors also thank Piero Rinaldo, MD, PhD, Mayo Clinic, for his assistance in the preparation of the figures and for granting permission to reproduce material from the R4S and CLIR websites.

References

The following references added:

- Gempel K, Kiechl S, Hofmann S et al (2002) Screening for carnitine palmitoyltransferase II deficiency by tandem mass spectrometry. *J Inherit Metab Dis* 25:17–27
- Hall PL, Marquardt G, McHugh DMS et al (2014) Post-analytical tools improve performance of newborn screening by tandem mass spectrometry. *Genet Med* 16:889–895

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