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Erratum

Erratum to "Infants Diagnosed with Spinal Muscular Atrophy and 4 *SMN2* Copies through Newborn Screening – Opportunity or Burden?"

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The Abstract has been corrected with the following bold text:

Abstract

Although the value of newborn screening (NBS) for early detection and treatment opportunity in SMA patients is generally accepted, there is still an ongoing discussion about the best strategy in children with 4 and more copies of the *SMN2* gene. This gene is known to be the most important but not the only disease modifier.
In SMA-NBS projects in Germany comprising 278,970 infants (200,901 screened between January 2018 and May 2019 in a pilot project initiated and recently published by the cystinosis foundation[1] and 78,069 screened in a subsequent project from June 2019 –November 2019) 38 positive cases with a homozygous *SMN*1 deletion were detected. 40% of them had 4 or more *SMN2* copies. The incidence for homozygous *SMN1* deletion was 1 : 7350, which is within the known range of SMA incidence in Germany.
Of the 15 SMA children with 4 *SMN2* copies, one child developed physical signs of SMA by the age of 8 months. Two children had affected siblings with SMA Type III, who were diagnosed only after detection of the index patient in the NBS. One had a positive family history with an affected aunt (onset of disease at the age of 3 years). Three families were lost to medical follow up; two because of socioeconomic reasons and one to avoid the psychological stress associated with the appointments.
Decisions on how to handle patients with 4 *SMN2* copies are discussed in the light of the experience gathered from our NBS pilot SMA **clinical study.**

[1]Hohenfellner K, Bergmann C, Fleige T, Janzen N, Burggraf S, Olgemöller B, et al. Molecular based newborn screening in Germany: Follow-up for cystinosis. Mol Genet Metab Rep. 2019;21 : 100514-.

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On page 2, second column, last paragraph of the Introduction section, the following sentence (bold text) has been updated:

In this paper, we discuss the limitations and the medical and ethical issues associated with the recommended procedure, based on our experience with a cohort of patients with 4 *SMN2* copies, who were detected **in the NBS projects** between January 2018 and November 2019. Specific problems are highlighted by 6 short case reports.

The Acknowledgments section has been corrected to the following text:

ACKNOWLEDGMENTS

The Cystinosis Foundation initiated, designed and conducted the pilot project for genetic newborn screening for SMA and cystinosis in Germany in 2017. Within this pilot project, in the period from January 2018 to May 2019, 200,901 newborns were tested and a total of 29 newborns with a homozygous deletion in the SMN1 gene were identified.[2]
The Cystinosis Foundation had no influence in interpretation and publication of the clinical data.

[2]Hohenfellner K, Bergmann C, Fleige T, Janzen N, Burggraf S, Olgemöller B, et al. Molecular based newborn screening in Germany: Follow-up for cystinosis. Mol Genet Metab Rep. 2019;21 : 100514-.