

Accepted Manuscript

Comprehensive Evaluation of Plasma 7-Ketocholesterol and Cholestan- $3\beta,5\alpha,6\beta$ -Triol in an Italian Cohort of Patients Affected by Niemann-Pick Disease due to *NPC1* and *SMPD1* Mutations

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PII: S0009-8981(16)30003-1  
DOI: doi: [10.1016/j.cca.2016.01.003](https://doi.org/10.1016/j.cca.2016.01.003)  
Reference: CCA 14236

To appear in: *Clinica Chimica Acta*

Received date: 23 October 2015  
Revised date: 24 November 2015  
Accepted date: 4 January 2016



Please cite this article as: Romanello Milena, Zampieri Stefania, Bortolotti Nadia, Deroma Laura, Sechi Annalisa, Fiumara Agata, Parini Rossella, Borroni Barbara, Brancati Francesco, Bruni Amalia, Russo Cinzia V., Bordugo Andrea, Bembi Bruno, Dardis Andrea, Comprehensive Evaluation of Plasma 7-Ketocholesterol and Cholestan- $3\beta,5\alpha,6\beta$ -Triol in an Italian Cohort of Patients Affected by Niemann-Pick Disease due to *NPC1* and *SMPD1* Mutations, *Clinica Chimica Acta* (2016), doi: [10.1016/j.cca.2016.01.003](https://doi.org/10.1016/j.cca.2016.01.003)

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# Comprehensive Evaluation of Plasma 7-Ketocholesterol and Cholestan-3 $\beta$ ,5 $\alpha$ ,6 $\beta$ -Triol in an Italian Cohort of Patients Affected by Niemann-Pick Disease Due to *NPC1* and *SMPD1* Mutations.

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## Abbreviations

3 $\beta$ ,5 $\alpha$ ,6 $\beta$ -triol: cholestan-3 $\beta$ ,5 $\alpha$ ,6 $\beta$ -triol; 3 $\beta$ ,5 $\alpha$ ,6 $\beta$ -triol-DMG<sub>2</sub>: bis-(dimethylglycinate)-derivative of 3 $\beta$ ,5 $\alpha$ ,6 $\beta$ -triol; 7-KC: 7-ketocholesterol; [<sup>2</sup>H<sub>7</sub>]-7-KC: 25,26,26,26,27,27,27-[<sup>2</sup>H<sub>7</sub>]-7-ketocholesterol; [<sup>2</sup>H<sub>7</sub>]-3 $\beta$ ,5 $\alpha$ ,6 $\beta$ -triol: 25,26,26,26,27,27,27-[<sup>2</sup>H<sub>7</sub>] cholestan-3 $\beta$ ,5 $\alpha$ ,6 $\beta$ -triol; APCI: atmospheric pressure chemical ionization; CE: collision energy; QC: quality control; CXP: collision cell exit potential; DMAP: 4-(dimethylamino) pyridine; DMG: dimethylglycine hydrochloride; DP: declustering potential; EDC: N-(3-dimethylaminopropyl)-N'-ethylcarbodiimide hydrochloride; EP: entrance potential; IQR: interquartile range; KC-DMG: mono-dimethylglycinate-derivative of 7-KC; LLOQ: lower limit of quantification; LOD: limit of detection; LQC, MQC, HQC: low, medium and high plasma quality control respectively; Methanol-d<sub>4</sub>: tetradeuteromethanol; TCA: trichloroacetic acid; ULOQ: upper limit of quantification

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