AIM OF THE STUDY
Liver transplantation (LTx) is increasingly utilized as therapeutic option for intoxication-type inborn errors of metabolism (IT-IEM).

However, little is known about the impact of LTx on health-related quality of life (HRQoL) at follow-up, so the aim of the study was to investigate by parents’ perspective the impact of liver transplantation (LTx) on HRQoL in patients with IT-IEM.

METHODS
This single-centre study involved parents of 30 subjects with IT-IEM (14 organic acidurias (OA), 8 Propionic, 6 Methylmalonic), 10 urea cycle disorders (UCD: 3 CPS, 2 OTC, 5 ASL), 6 maple syrup urine disease (MSUD) transplanted between 2011-2021 at Bambino Gesù Children’s Hospital (age at transplantation 6.8±7.3 years; range 8 months - 26 years; follow-up duration: 3.3±2.6 years; range 0.1-9 years). Parents completed two HRQoL questionnaire: the PedsQL 4.0TM as a generic HRQoL measure, assessed pre/post transplantation and, retrospectively, the MetabQoL 1.0TM, a tool recently developed to evaluating HRQoL in IT-IEM specifically.

RESULTS
QoL Total Score Pre-Post transplantation

By analysing separately, the three disease groups (OA, UCD, and MSUD), a significant improvement was observed in OA with both tools. However, by using MetabQoL Total Score, also UCD and MSUD patients showed a significant improvement of QoL after transplantation.

+ 20.5% PedsQL Total Scores improvement at post-tx in OA
+ 39.7% MetabQoL Total Scores improvement at post-tx in OA
+ 39.1% MetabQoL Total Scores improvement at post-tx in UCD
+ 38.6% MetabQoL Total Scores improvement at post-tx in MSUD

QoL according to IT-IEM subtypes

In the total population, post- vs pre-transplantation evaluation highlighted significant improvement in HRQoL proxy-report Total Scores assessed with both tools (PedsQL and MetabQoL).

+ 14.0% PedsQL Total Scores improvement at post-transplantation
+ 39.3% MetabQoL Total Scores improvement at post-transplantation

QoL according to Age at transplantation

Dividing the sample according to the age at transplantation, PedsQL post-LTx was significantly higher in patients younger than 3 years at LTx, while the evaluation with MetabQoL showed a significant improvement in both groups.

+ 14.1% PedsQL Total Scores improvement at post-tx in patients < 3 yrs
+ 41.4% MetabQoL Total Scores improvement at post-tx in patients < 3 yrs
+ 37.1% MetabQoL Total Scores improvement at post-tx in patients > 3 yrs

CONCLUSIONS
This study shows that liver transplantation has a positive impact on QoL in patients with IT-IEM. The importance of assessing the impact of transplantation on HRQoL, a meaningful outcome reflecting patient's wellbeing beyond exclusively medical measure, is recommended. The use of two different tools, showed that the specifically designed MetabQoL provided a more precise data interpretation. Considering the positive changes post transplantation detected from parents’ perspective, further studies on largest patients’ series are needed to explore also patient perspective. Our findings also highlight the promising use of the innovative and recently developed MetabQoL to evaluate in a more specific way HRQoL in IT-IEM.