Copper Histidinase Treatment for Menkes Disease (Kinky Hair Syndrome)

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Background

- Copper is an essential trace element required for human health.
- Menkes disease is an X-linked recessive disorder caused by mutations in the copper transporter gene, ATP7A. Patients with Menkes disease are born with impaired ability to absorb copper from their diet.
- Molecular defects include small deletions or insertions of DNA, nonsense mutations, null mutations or premature stop codons.

Methods

- The primary efficacy endpoint of overall survival was met. The analysis demonstrated statistically significant improvement in overall survival in subjects with Menkes disease treated with CuHis compared with untreated subjects (HC-ET) and historically treated subjects (HC-LT) (Figure 4). The primary efficacy endpoint of overall survival was met in the CuHis-ET cohort, and survival in the CuHis-LT cohort was statistically superior to HC-LT (Figure 5).

Results

- In the CuHis-ET cohort, hypoglycemia (12.5%) and infant respiratory distress syndrome (25.0%) were the most common systemic complications due to prematurity at baseline (Table 1). In the CuHis-LT cohort, hypoglycemia (4.7%) and head hypotonia (22.0%) were the most common systemic complications due to prematurity at baseline.
- In the CuHis-ET cohort, hypoglycemia (12.5%) and infant respiratory distress syndrome (25.0%) were the most common systemic complications due to prematurity at baseline (Table 1). In the CuHis-LT cohort, hypoglycemia (4.7%) and head hypotonia (22.0%) were the most common systemic complications due to prematurity at baseline.
- The CuHis-ET cohort had a mean overall survival time of 17.6 months (95% CI: 11.5–28.6), which was statistically superior to the HC-LT cohort (P = 0.0001).

Conclusions

- The CuHis-ET cohort had a mean overall survival time of 17.6 months (95% CI: 11.5–28.6), which was statistically superior to the HC-LT cohort (P = 0.0001).

References


Disclosures

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- The authors declare no conflicts of interest.

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