Treatment discovered for deadly childhood disease

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Researchers have discovered that a treatment involving enzyme replacement therapy dramatically reduces the risk of death in children with Pompe disease, a rare genetic disorder in which most children die before their first birthday. The disorder causes profound muscle weakness and heart and breathing problems and affects as many as one in 40,000 births. The study is published in the online edition of *Neurology*.

"This form of treatment has changed the natural history of this otherwise lethal disease," said study author Priya Sunil Kishnani, MD, with Duke University in Durham, North Carolina.

The year long study involved 18 children under the age of six months with rapidly progressing Pompe disease. Pompe disease is caused by a deficiency in the enzyme acid a-glucosidase (GAA), which is needed to break down glycogen, a complex sugar molecule which releases glucose.

The study found all 18 children who started to receive the enzyme replacement, recombinant human GAA (rhGAA), before they were six months old survived to at least 18 months of age. Fifteen of the 18 children also did not need a ventilator. The study showed that starting rhGAA before the age of six months reduced the risk of death in children by 99 percent, reduced the risk of death or invasive breathing assistance by 92 percent, and reduced the risk of death or any type of ventilation by 88 percent, compared to past patients without this treatment.

"This form of enzyme replacement therapy markedly extended survival and improved respiratory performance in these children, with a majority of them showing normal growth and substantial gains in motor development," said Kishnani. "rhGAA is safe and the only effective treatment for Pompe disease; it is life saving."

Kishnani said the young age at which the children began treatment may have contributed to their improved response compared to previous trials with rhGAA, where patients were older.

"This study demonstrates that starting enzyme replacement therapy early, which could be facilitated by newborn screening, shows great promise to reduce the mortality and disability of babies with this devastating disorder," said Kishnani.

The most common side effects of the rhGAA treatment included skin reactions such as rash and hives, fever, and changes in heart rate. The study was supported by the Genzyme Corporation, maker of rhGAA.

Source: American Academy of Neurology