Expanded genetic testing of newborns could help us get the most out of new gene therapies

The company behind [spinal muscular atrophy drug] AveXis, is seeing the most dramatic results in children who are treated in the first month or two of life.

That means identifying patients as soon as possible—ideally at birth. Finding them early could mean the difference between life and death. But as AveXis and other biotech companies race to bring transformative gene therapies to market, states have been slow to adopt screening tests for genetic diseases that are, increasingly, treatable.

Newborn screening tests in the US cover a minimum of 34 disorders, and many states have opted to add more. Yet most don't look for spinal muscular atrophy (commonly abbreviated as SMA), even though it's the leading genetic cause of infant death.

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Other gene therapies may also work better in children before a genetic defect has time to irreparably damage the body. For example, Bluebird Bio is developing one that halted a deadly brain disorder called cerebral adrenoleukodystrophy (ALD), also known as Lorenzo's Oil disease, in 15 out of 17 children. In a statement provided to MIT Technology Review, the company said outcomes are better when patients are treated before symptoms appear.

On February 8, a national committee that oversees newborn testing voted to recommend that SMA be added to the recommended universal screening panel.

Read full, original post: Gene therapy is saving children's lives—but screening to discover who needs it is lagging behind