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Using gene therapy and metabolite supplementation to treat ciliopathies

Stanford scientists have discovered that novel gene therapy and metabolite supplementation approaches show promise in treating ciliopathies. These potential breakthroughs could significantly impact patients, as there are currently no approved treatments for any ciliopathy condition.

Ciliopathies are genetic disorders affecting multiple organ systems and reducing life expectancy. Joubert syndrome, a rare form of ciliopathy, is characterized by midbrain defects, ocular motor apraxia, and developmental delays. At least 35 genes critical for cilia function have been identified in these disorders. Ciliopathies can lead to renal failure, progressive blindness, and other severe complications. Despite the identification of key genes like ARMC9 and the significant impact on patients' quality of life, there are currently no approved treatments for any ciliopathy condition, highlighting the urgent need for novel therapeutic approaches.

Gene therapy and metabolite supplementation resulted in a rescue of cilia formation in Joubert Syndrome patient cells. Importantly, these treatments rescued cellular defects and cilia deficiency *in vivo* and in cellular models. Consequently, gene therapy and metabolite supplementation have the potential to significantly improve patient outcomes and transform the treatment landscape for ciliopathies by addressing the fundamental cellular and molecular defects underlying these disorders.

Stage of Development
Preclinical - in vivo
Continued research - continuing with future applications for NIH funding

Applications

- Treatment of ciliopathies, including Joubert Syndrome

- Addressing cellular defects and cilia formation in genetic disorders
- Potential therapy for multiple organ systems affected by ciliopathies

Advantages

- Gene therapy rescues cilia formation in Joubert Syndrome patients
- Metabolite supplementation restores cellular defects and cilia deficiency *in vivo* and in cells
- Versatile approaches that can potentially address multiple ciliopathy manifestations

Innovators

- Yang Sun
- Chien Hui Lo

Licensing Contact

Hyunjin Kim

Licensing Manager, Life Sciences

Email