GENE EDITING THERAPY: SCIENCE & TECHNOLOGY

NEWS: World's first gene-editing therapy saves a baby with rare disorder: Why is this significant?

WHAT'S IN THE NEWS?

In a historic first, scientists in the US have successfully used CRISPR-based gene-editing therapy to treat an infant with Carbamoyl Phosphate Synthetase 1 (CPS1) deficiency—a rare genetic disorder. This marks a major breakthrough in personalized medicine, though it raises safety, ethical, and access-related concerns.

Historic Medical Breakthrough: CPS1 Gene-Editing Therapy

- In 2024, scientists in the US successfully used a personalized CRISPR-based gene-editing therapy to treat a rare genetic condition — Carbamoyl Phosphate Synthetase 1 (CPS1) deficiency — in an infant.
- This marks the first instance of using CRISPR gene therapy in humans to address this metabolic disorder.

Understanding CPS1 Deficiency

- It is a rare autosomal recessive genetic disorder.
- The liver lacks the enzyme Carbamoyl Phosphate Synthetase 1, which is essential for the urea cycle — the process that converts toxic ammonia into excretable urea.
- Without treatment, toxic ammonia accumulates in the blood, leading to neurological damage, coma, or death, especially in infants.

What is Gene Editing Therapy?

- It is a medical technique that involves altering DNA within a person's cells to correct genetic defects or introduce therapeutic changes.
- Targets diseases caused by specific gene mutations using precision tools to repair or modify DNA.

Types of Gene Editing Techniques

- CRISPR-Cas9: Uses a guide RNA and Cas9 enzyme to cut DNA at specific locations.
- Zinc Finger Nucleases (ZFNs): Uses engineered proteins to bind and cleave DNA at desired spots.

- Base Editing: Alters a single nucleotide base pair without breaking the DNA strand.
- Prime Editing: A sophisticated method that can “search and replace” DNA bases with high precision.

How CRISPR Works (Mechanism)

- Identify the faulty gene.
- Create a guide RNA that matches the mutation.
- Cas9 enzyme, guided by RNA, cuts the defective DNA at the target site.
- The cell’s natural repair mechanism inserts the correct gene sequence.

Applications of Gene Editing

- Human Medicine:
    - Treats monogenic diseases like Sickle Cell Anaemia, Thalassemia, and CPS1 deficiency.
    - Cancer therapy (e.g., CAR-T cell therapy).
- Agriculture:
    - Produces pest-resistant, drought-tolerant, and high-nutrition crops.
- Veterinary Science:
    - Enhances productivity, disease resistance in livestock.

India’s Progress in Gene Editing

- IndiCRISPR: An indigenous CRISPR platform developed by CSIR-IGIB.
- National Biopharma Mission: Supports genome-editing research under DBT.
- Gene Therapy Guidelines (2020):
    - Laid out by ICMR and DBT.
    - Focuses on ethics, safety, and clinical trials for gene therapies in India.

Challenges in Gene Editing

- Safety Concerns: Risk of unintended edits (off-target effects), immune reactions, and long-term consequences.
- Ethical Dilemmas:
    - Germline editing can cause permanent heritable changes.
    - Risk of “designer babies” raises socio-ethical concerns.
- Equity & Access:
    - Gene therapy remains costly and largely confined to high-income countries.
    - Infrastructure for large-scale delivery and regulation is still developing.

Conclusion

Gene editing therapy, especially CRISPR-based, represents a revolutionary advancement in personalized medicine. The success in treating CPS1 deficiency signals the start of a new era in gene-based therapies, but it also brings regulatory, ethical, and accessibility challenges that must be addressed responsibly.

Source: https://indianexpress.com/article/health-wellness/world-first-gene-editing-therapy-saves-baby-rare-disorder-significant-10010295/