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REVIEW ARTICLE

Contribution of Jennifer Doudna in the Field of Pediatrics

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ABSTRACT

Jennifer Doudna's scientific journey transformed modern pediatrics by pioneering CRISPR-Cas9, a genome-editing technology that has reshaped the diagnosis, prevention, and treatment of genetic disorders. Beginning with her early fascination for biology and rigorous training in molecular biology and RNA research, Doudna's collaboration with Emmanuelle Charpentier led to the discovery of the CRISPR-Cas9 editing mechanism, marking a revolutionary breakthrough. Their work enabled precise, efficient, and accessible gene editing, opening therapeutic possibilities for conditions such as sickle cell disease, thalassemia, Leber congenital amaurosis, Duchenne muscular dystrophy, and several neurodegenerative disorders. CRISPR also advanced prenatal diagnostics and contributed to rapid vaccine development during the COVID-19 pandemic. Despite its power, the technology raised significant ethical concerns, including the controversial creation of "designer babies," prompting global discussions on responsible use. Awarded the 2020 Nobel Prize in Chemistry, Doudna's work symbolizes innovation, perseverance, and the transformative potential of science in shaping pediatric healthcare.

Key Message: CRISPR-Cas9, co-developed by Jennifer Doudna, represents a landmark innovation that is reshaping pediatric healthcare by enabling targeted correction of disease-causing genes. As its therapeutic applications expand, responsible scientific stewardship and ethical oversight remain essential to ensuring that the technology is used to alleviate human disease while safeguarding future generations.

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INTRODUCTION

"This was a new room, rich with hope, terrible with strange danger. A dim folk memory had preserved the story of a greater advance: "the winged hound of Zeus" tearing from Prometheus' liver the price of fire. Was the world ready for the new step forward? Certainly, it will change the world. You have to make laws to fit it. And if plain people did not understand and control it, who would?"

The revolution that commenced around a century ago is rather focussed on the three fundamental notions of our very existence: the atom (relativity and quantum mechanics), the bit (computers and internet) and the gene (the genetic coding).

Everything revolves around The Code.

Jennifer Doudna with her co-partner Emmanuelle Charpentier, in 2020, won the Nobel Prize in Chemistry which was awarded to them for Developing a method of Genome Editing – the CRISPRCas9 technology.

It is an established fact that the society has certain limitations imposed upon women, the scientific world hence is no different. In the world which screams of male chauvinism, this is a small insight in the breakthrough journey of Jennifer Doudna, who faced her fair share of challenges to come up with the gene editing tool with the help of her co-partner. Both, Jennifer and Emmanuelle are the part of the only female duo in the history to ever share a Nobel Prize without any man's name in it. Hence, the scientist Jennifer Doudna has been chosen.

A glimpse of what is she made of:

- *Early life:* Jennifer was born on 19 February 1964, to Martin Doudna, a professor of English Literature and Dorothy Doudna, a history teacher; in Washington DC. She grew up in Hilo, Hawaii.

  As a child, eyes always bubbling with curiosity and soul scintillating with desire to learn, her father played an immense role in shaping her persona and building her intellect. He introduced her to Science through various books, the most notable of it all, *The Double Helix* by James Watson, which inspired her to make a career in biochemistry and genetics. The beautiful atmosphere of Hilo, the myriads of vegetation and small creatures there embarked her fascination with biology too. When she knew that Science was what she was born to do – the isolated environment and the people from her high school and college often demotivated her from pursuing the field. Her little world there told her that women don't do science, it's an area of men's expertise and excellence. That was her cue to leave Hilo and going ahead away from home for higher education.
- *Education:* She studied chemistry at Pomona College in California. Then pursued graduate work at Harvard, under the guidance of Jack Szostak, for molecular biology. One day, Doudna was experimenting with yeast. Yeast cells are efficient in taking up pieces of DNA and integrating them into their genetic makeup. She used this knowledge, with a little bit of electric spark, succeeded in wriggling her lab made DNA sequence (which matched with the yeast's) into the yeast's DNA.

  She realised she had made a little tool that could edit genes. This is where it all began.
- *Personal experiences:* It was roughly the time when the human genome was coded down in the Human Genome Project. Doudna learnt about it but realised that having a map of DNA was not a breakthrough because the mutations in them still existed giving rise to Sickle cell or Huntington's. She then found her interest shifting towards RNA, the worker bee which actually makes different set of tools for DNA, the molecule which had catalytic powers of enzymes. She worked hard to delineate the structure of RNA using X-ray crystallography (which was

an accident). She then focussed on making Ribozyme in the lab with Szostak's help. Eventually they engineered a ribozyme that could splice together a copy of itself. They also found out about the repeat sequence in bacteria which was similar to that of the virus that infected that particular bacteria. This made them understand that bacteria developed a memory against the same virus. She was not the only one trying to work on this process of finding a tool to edit genes.

The first researcher to figure out the function of repeated sequence was Francisco Mojica, a graduate student at the University of Alicante, Spain. He called these repeat sequence CRISPR, for "clustered regularly interspaced short palindromic repeats." These CRISPR sequence encoded directions for making an enzyme which he called "CRISPR associated," or *Cas* enzyme. He then published this finding. Doudna then came to know about the fact that other people did know about it and gave it a name too. Even though the sequence was named, nobody knew how the bacteria used it to protect itself from virus' infection. She then moved to Berkeley to work upon it in her own lab. She, with her research partner Martin Jinek, discovered that Cas1 has a distinct fold, indicating that it is the mechanism that bacteria uses to cleave a snippet of DNA from invading viruses and incorporate it into their CRISPR array, thus being key to the memory forming system. Doudna had discovered the components required to work the gene editing in the test tube but it was lacking. She had two things – CRISPR and Cas9 enzyme (this was the vital enzyme, more important than Cas1) but she needed the third element to perform the editing. Then came in Emmanuelle Charpentier.

Emmanuelle, a French biologist, too had been studying CRISPR and homed in on the enzyme- Cas9. She discovered the 'third element' – the "trans-activating CRISPR RNA" or tracrRNA. Together with Doudna, they were successful in making the first successful gene editing experiment possible in a test tube. That made this duo powerful because even though there were many people experimenting with CRISPR, none of them could discover Cas9. Then they published a paper in 2012, which showed that this bacterial system could be adapted into a revolutionary gene-editing tool for scientists.

The difference it made, the power CRISPR has:

- *Scientific Contribution:* To elaborate further on how this CRISPR techniques was revolutionary, we need to understand how it works.

This tool, once granted the patent for, was used in treating genetic conditions.

- *Curing sickle cell disease:* as this was the simplest point mutation, Doudna's students started using CRISPR to treat the genetic defect. This mutation causes RBCs to become sickleshaped, leading to pain, organ damage and early death. The aim was to turn off a gene that suppresses fetal hemoglobin which would in turn reactivate the healthy hemoglobin which could replace the faulty one. The first patient in trial was Victoria Gray, a mother of four children. The doctors first took her stem cells out to edit the faulty gene and to activate the gene that suppresses fetal hemoglobin. Then they injected this into Victoria. Ever since she got the edited cells, she hadn't needed to get donor transfusions or had any attacks of pain. After nine months, the percentage of fetal hemoglobin increased to a total of whopping 81%.
- *Curing Thalassemia:* the aim here was to again boost the fetal hemoglobin cells to compensate for the insufficiency rooted because of the disorder. Several patients have been freed from regular blood transfusions.
- *Way to gift sunshine to the blinds:* the goal was to treat Leber Congenital Amaurosis (LCA) which is a common cause of childhood blindness. Those with the condition have a mutation in the gene that makes light – receptor cells in their eye which causes error in the light signals to get converted into nerve signals. Direct injection of CRISPR into the eye beneath the retina was given. Trials showed partial restoration of vision in the patients.
- *Hopes to restore muscle, guiding children's way to movements:* experimental therapies were tried for Duchenne Muscular Dystrophy which is a severe muscle wasting disease that begins early in life and leads to loss of mobility and early death. Aim was to restore the dystrophin

protein by correcting mutations. Preclincial studies show hope for slowing and even reversing the muscle damage in children

- *Making the child's future better before the child is born:* CRISPR was widely used for prenatal diagnosis and correction of lasting genetic conditions as a result of which, the life to be born was ensured a disease free survival.
- *A holistic tool not only for the young but for the whole world:* CRISPR was and also tried for diseases like Huntington's Chorea, Alzheimer's, Cancers like lung cancer, and in the COVID pandemic. It was because of CRISPR the Pfizer vaccine was made against the Corona Virus.

The thorns and stones that blocked the way:

- *Challenges faced:* when a tool this powerful is out in the open for the mass to use, it ensures attracting the ethical and moral issues. The case with CRISPR was no different. Doudna was elated to have this revolutionary breakthrough be used by the doctors to treat life hampering diseases but not everyone was on the same page as hers.

It gave a plethora of possibilities how about we edit the colour of our eyes? How about we increase the IQ of our child? How about we enhance our immunity? How about we makes our muscle grow very bulky and strong? How about we edit our characters? What does it mean for humanity? Should we be hacking our own DNA to make us less susceptible to disease? Should we democratize the technology that would allow parents to enhance their kids?

The Designer Babies: CRISPR used in a human experiment format by Chinese scientist He Jiankui in 2018, to edit the embryo prenatally, which could create inheritable changes. He edited the embryo of the child who was to be born to HIV infected mother and father. He edited the CCR-5 gene and the mother actually gave birth to HIV free twins who were known as the designer babies. The issue was ethical because nobody should have power to do this as an experiment. A red line was then drawn after many meetings between officials worldwide including Doudna that germline gene editing must be prohibited. Norms were made and imparted upon the people to follow.

Doudna knew that everything had two sides. "Someday we may consider it unethical *not* to use germline editing to alleviate human suffering. As long as we are correcting genetic mutations by restoring the 'normal' version of the gene, we're likely to be on the safe side. Science doesn't move backwards, and we can't unlearn this knowledge, so we need to find a prudent path forward. We have now the power to control our genetic future, which is awesome and terrifying. So we must move forward cautiously and with respect for the power we've gained", said Doudna in 2015 Napa Valley meeting.

"Men ought not to play God before they learn to be men" – Fabricated Man by Paul Ramsey.

The final breakthrough, the moment this world witnessed history:

- Rewriting the Code of Life: NOBEL PRIZE 2020: Doudna was asleep when, at 2:53 AM, on October 9, 2020, she was awakened by the persistent buzz of her phone. The call was from a reporter for Nature. "I hate to bother you so early," she said, "but I wanted to comment on the Nobel. You and Emmanuelle Charpentier won!"

This prize was given to them about rewriting the code of life. History was made, prize went to two women without a man involved for the very first time. One could sense justice for the women in the past who were owned by masculinity. The women who tried but couldn't have their breakthrough. Women like Rosalind Franklin, who even after discovering DNA, didn't get credit because of the man who inspired Doudna's journey, James Watson. Such is the irony of time. Doudna's struggles and challenges were answered. Finally, the code was broken.

The plant of change it watered in me:

The one lesson I could imbibe from this very beautiful journey is – Perseverance. Doudna's journey was inked on the pages made of perseverance and patience. She was such a humble lady, such a modest colleague to everyone around her, such an intelligent lady who let the spark of 'I want to belong, I want to create' burn until it turned bright enough for the world to see. There's only little to do says the mind who knows little, reading about Doudna will definitely help me widen my

horizons, to set new limits much higher than I could ever imagine for myself.

The second lesson I would love to inculcate in me would be to never forget the fact if my success is a plant upright, the roots are made of failures. Failures are an inseparable part of any journey, so to learn to accept it, to learn from it and grow will be the intention henceforth.

The whole excerpt was based on the book *The Code Breaker* written by *Walter Isaacson*. This book made me understand Doudna very personally. It was written in a way to make every reader feel as if it's the story about them.

CONCLUSION

CRISPR-Cas9, co-developed by Jennifer Doudna, represents a landmark innovation that is reshaping pediatric healthcare by enabling targeted correction of disease-causing genes. As its therapeutic applications expand, responsible scientific stewardship and ethical oversight remain essential to ensuring that the technology is used to alleviate human disease while safeguarding future generations.

REFERENCES

1. The Code Breaker by Walter Isaacson, a novel.
2. Foundational CRISPR-Cas9 Discovery Papers - Jinek *et al*., 2012 – The CRISPR–Cas9 Breakthrough Paper, "A Programmable Dual-RNA–Guided DNA Endonuclease in Adaptive Bacterial Immunity."
3. Charpentier *et al*., 2011 – Discovery of tracrRNA - "tracrRNA and CRISPR/Cas system in Streptococcus pyogenes"
4. Deltcheva *et al*., 2011 – RNA Processing in CRISPR - "CRISPR RNA maturation by RNase III"
5. Mojica *et al*., 2003–2005 – Identification of CRISPR as an immune system - "Short clustered repeats… in prokaryotes"
6. Cong *et al*., 2013 – First use of CRISPR in human cells (Broad Institute) - "Multiplex Genome Engineering Using CRISPR/Cas Systems"
7. Barrangou *et al*., 2007 – Proof that CRISPR is immunity - "CRISPR Provides Acquired Resistance Against Viruses in Prokaryotes"
8. Gootenberg *et al*., 2017 – SHERLOCK Diagnostic CRISPR Tool - "Nucleic acid detection using CRISPR enzymes"
9. He Jiankui CRISPR Baby Papers (2018)
10. Doudna's early ribozyme and RNA folding papers (1990s–2000s)