



DNA

Scientists have found the probable cause of the IMAGE syndrome, which is responsible for restricted growth in babies.

This research has been published online in the May 27 issue of the journal *Nature Genetics*.

IMAGE syndrome is a kind of puzzle for scientists for over 20 years. Babies with this syndrome have stunted growth before birth and cause the smaller-than-normal body and organs. It can be life-threatening in some situations.

Eric Vilain, study research from University of California, Los Angeles (UCLA) first detected the disease in his native country France, where he was caring for two boys, who were 3 and 6 years of age and who were very short of their ages.

“I never found a reason to explain these patients’ unusual set of symptoms,” Vilain said in a statement. “I’ve been searching for the cause of their disease since 1993.”

In this study, researchers worked on patients with IMAGE syndrome from around the world and assayed them genetically. They compared the genomes of the patients with healthy family members and found a mutation in a gene called as CDKN1C.

The mutation in the CDKN1C gene “consistently appeared in every family member affected by IMAGE syndrome,” Vilain said in a statement. “We were a little surprised, though, because the mutation was located on a gene previously recognized as causing Beckwith-Wiedemann syndrome.”

Beckwith-Wiedemann syndrome is a growth disorder related to large body sizes and organs.

“Finding dual functions in one molecule is an unusual biological phenomenon. These two diseases are polar opposites of each other, Vilain said. “When the mutation appeared in the slim section we identified, the infant developed IMAGE syndrome. If the mutation fell anywhere else in the gene, the child was born with Beckwith-Wiedemann. That’s really quite remarkable.”

Researchers are optimistic that this discovery could help in developing therapeutic strategy for the stunted growth and its related problems.

Reference:

Valerie A Arboleda, Hane Lee, Rahul Parnaik, Alice Fleming, Abhik Banerjee, Bruno Ferraz-de-Souza, Emmanuèle C Délot, Imilce A Rodriguez-Fernandez, Debora Braslavsky, Ignacio Bergadá, Esteban C Dell’Angelica, Stanley F Nelson, Julian A Martinez-Agosto, John C Achermann & Eric Vilain, (2012). Mutations in the PCNA-binding domain of CDKN1C cause IMAGE syndrome. *Nature Genetics*, doi:10.1038/ng.2275

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