



National Human Genome Research Institute

Mouse Model of Diamond-Blackfan Anemia

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KEY WORDS

Diamond-Blackfan Anemia, DBA, RPS7, Ribosomal Proteins, Mouse Model

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PATENT-PENDING TECHNOLOGY AVAILABLE FOR LICENSING

SUMMARY

Diamond-Blackfan Anemia (DBA) is a ribosomal protein disease that results in reducing the level of red blood cells. Ribosomes translate mRNA into protein and consist of two subunits, each composed of a large number of proteins, including small ribosomal proteins (RPSs) and large ribosomal proteins (RPLs). Mutations in ribosomal proteins (i.e., RPL5, RPL11, RPL35A, RPS7, RPS10, RPS17, RPS19, RPS24 and RPS26) have been found to occur in 50-60% of DBA patients. Investigators at the National Human Genome Research Institute (NHGRI) showed that mutations in RPS7 (found in about 1% of DBA patients) cause disruption of ribosome biogenesis, as well as abnormal skeletal, melanocyte and central nervous system development. They generated Zuma mutant mouse line (RPS7Zma) with an A to C point mutation in exon 7 of Rps7 predicted to cause substitution of a conserved amino acid. These mice show abnormal ribosomal biogenesis and deficits in working memory, reduced melanocyte development, and underdeveloped cortex.

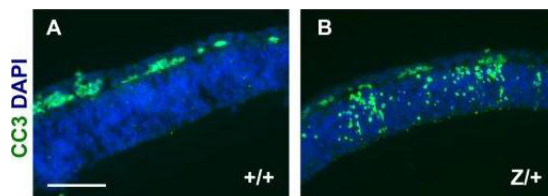
POTENTIAL COMMERCIAL APPLICATIONS

The described RPS7Zma can be used to further understand the DBA disease. It can, however, also be a tool for investigating other defects of mammalian development (e.g., the development of the central nervous system).

RELATED ARTICLES

Watkins-Chow et al., "Mutation of the Diamond-Blackfan anemia gene Rps7 in mouse results in morphological and neuroanatomical phenotypes," PLoS Genetics 2013 Jan;9(1):e1003094. Epub 2013 Jan 31.

<http://www.plosgenetics.org/article/info%3Adoi%2F10.1371%2Fjournal.pgen.1003094>



Increased apoptosis was observed in coronal sections through the neocortex on embryonic day 11.5 in the Zuma model (Z/+) compared to wild type RPS7 (+/+).

