POLR3B Gene

Subjects: Genetics & Heredity
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RNA polymerase III subunit B

genes

1. Introduction

The *POLR3B* gene provides instructions for making one part (subunit) of an enzyme called RNA polymerase III. This enzyme is involved in the production (synthesis) of ribonucleic acid (RNA), a chemical cousin of DNA. The RNA polymerase III enzyme attaches (binds) to DNA and synthesizes RNA in accordance with the instructions carried by the DNA, a process called transcription. RNA polymerase III helps synthesize several forms of RNA, including ribosomal RNA (rRNA) and transfer RNA (tRNA). Molecules of rRNA and tRNA assemble protein building blocks (amino acids) into working proteins; this process is essential for the normal functioning and survival of cells.

2. Health Conditions Related to Genetic Changes

2.1. Pol III-related leukodystrophy

At least 54 *POLR3B* gene mutations have been associated with Pol III-related leukodystrophy. Leukodystrophies are conditions that involve abnormalities of the nervous system's white matter. White matter consists of nerve fibers covered by a fatty substance called myelin, which insulates nerve fibers and promotes the rapid transmission of nerve impulses. A reduced ability to form myelin (hypomyelination) leads to the signs and symptoms of Pol III-related leukodystrophy, which include intellectual disability and difficulty with coordinating movements (ataxia). Development of the teeth (dentition) is also abnormal in this disorder.

In the Pol III-related leukodystrophies, *POLR3B* gene mutations may impair the ability of the subunits of the RNA polymerase III enzyme to assemble properly or result in an RNA polymerase III with impaired ability to bind to DNA. Reduced function of the RNA polymerase III molecule likely affects development and function of many parts of the body, but the relationship between *POLR3B* gene mutations and the specific signs and symptoms of these disorders is unknown.

People with Pol III-related leukodystrophy may have different combinations of its signs and symptoms. These varied combinations of clinical features were originally described as separate disorders. Affected individuals may be diagnosed with ataxia, delayed dentition, and hypomyelination (ADDH); hypomyelination, hypodontia,

hypogonadotropic hypogonadism (4H syndrome); tremor-ataxia with central hypomyelination (TACH); leukodystrophy with oligodontia (LO); or hypomyelination with cerebellar atrophy and hypoplasia of the corpus callosum (HCAHC). Because these disorders were later found to have the same genetic cause, researchers now group them as variations of the single condition Pol III-related leukodystrophy.

3. Other Names for This Gene

- C128
- DNA-directed RNA polymerase III 127.6 kDa polypeptide
- DNA-directed RNA polymerase III subunit B
- DNA-directed RNA polymerase III subunit RPC2
- DNA-directed RNA polymerase III subunit RPC2 isoform 1
- DNA-directed RNA polymerase III subunit RPC2 isoform 2
- FLJ10388
- HLD8
- polymerase (RNA) III (DNA directed) polypeptide B
- polymerase (RNA) III subunit B
- RNA polymerase III subunit C2
- RPC2
- RPC2_HUMAN

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