CARRIER STATUS REPORT


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LifeandMe مفتخر است شما را به دنیایی دعوت کند که از لحظه تولد همراه شما است و از این پس امکان جستجو در آن برایتان ممکن شده است. ژن‌های بدن تعیین‌کننده بسیاری از عوامل حیاتی در طول زندگی هستند که با شناخت بهتر آن‌ها می‌توان زندگی سالم‌تر و طول عمر بیش‌تری را به ارمغان آورد.

انسان‌ها در کلیات ژنتیکی با هم تشابه دارند؛ اما هیچ دو انسانی با هم برابر نیستند. وجود این تفاوت‌ها امکان شخصی‌سازی سبک زندگی را به ما می‌دهد که تخصص **LifeandMe** است. در ادامه این راه هیجان انگیز، شما به همراهی ما ژن‌ها را خواهید شناخت و راهکارهای شخصی‌سازی سبک زندگی خود را درخواهید یافت.

این گـــــزارش منحصـــراً بـــرای شـــخص شمـــا تولـید شـــده است.

ARSACS

Also known as: Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay

ARSACS is a rare genetic disorder characterized by loss of sensation and muscle control, as well as muscle stiffness that worsens over time. A person must have two variants in the SACS gene in order to have this condition.

MECHANISM

ARSACS is caused by variants in the **SACS** gene.

The SACS gene contains instructions for making a protein called sacsin, a protein of unknown function that is mainly present in the brain, skin cells, and skeletal muscles. Certain variants in SACS lead to a shortened protein that cannot function properly.

ETHNICITIES MOST AFFECTED

This condition is most common in people of French Canadian descent, particularly from the Charlevoix and Saguenay-Lac-Saint-Jean regions of Quebec.

STATUS

gene	rs number	your variant
SACS	rs281865117	Not Detected

REFERENCES

No. 1 to No. 6
from the final references list.

Agenesis of the Corpus Callosum with Peripheral Neuropathy

02

Also known as: Andermann Syndrome

ACCPN is a rare genetic disorder. It is characterized by an incomplete connection between the two sides of the brain. This causes developmental disability, weakness, and loss of sensation. A person must have two variants in the SLC12A6 gene in order to have this condition.

ETHNICITIES MOST AFFECTED

This condition is most common in people of French Canadian descent, particularly from the Charlevoix and Saguenay-Lac-Saint-Jean regions of Quebec.

STATUS

gene	rs number	your variant
SLC12A6	rs515726215	Not Detected

MECHANISM

ACCPN is caused by variants in the **SLC12A6** gene.

The SLC12A6 gene contains instructions for making a protein called potassium-chloride (K-Cl) cotransporter. This protein controls the levels of water, potassium, and chloride inside of cells, which is important for proper nerve function. Certain variants in SLC12A6 disrupt this protein's function, leading to altered brain development and activity.

REFERENCES

No. 7 to No. 11
from the final references list.

05

Autosomal Recessive Polycystic Kidney Disease

ARPKD is a rare genetic disorder. It is characterized by kidney, liver, and lung problems as well as urinary tract infections and high blood pressure. A person must have two variants in the PKHD1 gene in order to have this condition.

MECHANISM

ARPKD is caused by variants in the **PKHD1** gene.

The PKHD1 gene contains instructions for making a protein called fibrocystin that is primarily found in the kidneys. Although its exact function is unknown, it is thought to play an important role in the development and function of the kidneys. Certain variants in PKHD1 disrupt its function.

ETHNICITIES MOST AFFECTED

This condition occurs in people of all ethnicities, but is best studied in people of Finnish, European, Hispanic, Turkish, and Middle Eastern descent.

STATUS

gene	rs number	your variant
PKHD1	rs137852949	Not Detected
	rs398124502	Not Detected

REFERENCES

No. 12 to No. 22
from the final references list.

Beta Thalassemia and Related Hemoglobinopathies

04

Also known as: Cooley's Anemia, Mediterranean Anemia

Beta thalassemia is a genetic disorder characterized by anemia and fatigue as well as bone deformities and organ problems. A person must have two variants in the HBB gene in order to have this condition.

ETHNICITIES MOST AFFECTED

This condition is most common in people of Mediterranean, Middle Eastern, North African, Transcaucasian, Central Asian, South Asian, Southeast Asian, and Southern Chinese descent.

STATUS

gene	rs number	your variant
HBB	rs34598529	Not Detected
	rs33960103	Not Detected
	rs33915217	Not Detected
	rs35724775	Not Detected
	rs35004220	Not Detected
	rs34451549	Not Detected
	rs34690599	Not Detected
	rs63750783	Not Detected
	rs11549407	Not Detected

MECHANISM

Beta thalassemia and related hemoglobinopathies are caused by variants in the **HBB** gene.

The HBB gene contains instructions for making a protein called beta-globin. This protein is part of a larger protein called hemoglobin that is found in red blood cells. Hemoglobin transports oxygen from the lungs to all other cells of the body. Certain variants in HBB alter the structure of hemoglobin, making it defective in transporting oxygen.

REFERENCES

No. 23 to No. 64
from the final references list.

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