PGP10 follow-up trait survey (June 2011)

This is a brief survey asking about some unusual genetic traits which might be predicted in one or more of the PGP10 individuals. We believe these are almost all false positives, but we would like to check with you to confirm each one. We may follow up with you if one of your answers appears to be consistent with a predicted phenotype.

As always, please do NOT use our findings as a medical test. This is for research purposes only.

Identification *

Please select which participant you are:

- PGP1 / George Church / hu438603
- PGP2 / John Halamka / huC30901
- PGP3 / Esther Dyson / huBEDA0B
- PGP4 / Misha Angrist / huE80E3D
- PGP5 / Kirk Maxey / hu9385BA
- PGP6 / Steven Pinker / hu04FD18
- PGP7 / Keith Batchelder / hu0D879F
- PGP8 / Stanley Lapidus / huAE6220
- PGP9 / Rosalynn Gill / hu034DB1
- PGP10 / James Sherley / hu604D39
- Other…

Kidney stones - individual

Have you ever had kidney stones?

- No
- Yes, once
- Yes, multiple times
- Other…

Kidney stones - relatives

Have any of your first degree relatives (parents, siblings, or children) had kidney stones?

- No / not that I am aware of
- Yes, one incident
- Yes, multiple incidents
- Other…

Polycystic kidney disease

Have you or a relative been diagnosed with polycystic kidney disease?

- No / not that I am aware of
- Yes
- Other…

- Yes
- Other...

Deafness

Do you have profound hearing loss / deafness or use hearing aids?

- No
- Yes
- Other...

Congenital heart defect

Have you or a relative had supravalvular aortic stenosis, a congenital heart defect?

- No / not that I am aware of
- Yes
- Other...

Heart disease: long-QT syndrome

Have you or a relative been diagnosed with long-QT syndrome?

- No / not that I am aware of
- Yes
- Other...

Heart disease: sudden death

Do you have a relative who has died suddenly due to cardiac failure at an unusually young age?

- No
- Yes
- Other...

Heart disease: hypertrophic cardiomyopathy

Have you or a relative been diagnosed with hypertrophic cardiomyopathy?

- No / not that I am aware of
- Yes
- Other...

Heart disease: cardiovascular disease

Have you or a first-degree relative been diagnosed with cardiovascular disease before the age of 50?

- No / not that I am aware of
- Yes, I have
- Yes, I have not but a first degree relative (parent, sibling, or child) has
- Other...

- Yes, I have
- Yes, I have not but a first degree relative (parent, sibling, or child) has
- Other…

Hypocholesterolemia

Have you or a relative been diagnosed with hypocholesterolemia (abnormally low cholesterol)?

- No / not that I am aware of
- Yes
- Other…

Amyloidosis

Have you or a relative been diagnosed with amyloidosis?

- No / not that I am aware of
- Yes
- Other…

Cutis laxa

Cutis laxa is a rare disorder in which the skin becomes inelastic and hangs loosely in folds. Do you or a relative have cutis laxa or similar symptoms?

- No / not that I am aware of
- Maybe, I or a relative has similar symptoms but has never been formally diagnosed.
- Yes, I or a relative has been diagnosed with cutis laxa.
- Other…

Palmar hyperlinearity

Some subtle skin phenotypes can be cause by heterozygous variants would would cause severe skin disorder if homozygous. These can include palmar hyperlinearity (causing a hand to look unusually old). Do you have palmar

- No
- Maybe
- Yes
- Other…

Keratosis pilaris

Some subtle skin phenotypes can be cause by heterozygous variants would would cause severe skin disorder if homozygous. These can include keratosis pelaris (bumps on the skin on the upper arms, cheeks, or thighs), or fine scale on the skin. Do you have keratosis pilaris?

- No
- Maybe
- Yes
- Other…

Cafe au lait spots

Do you have cafe au lait spots (light brown birthmarks)? If so, please describe how many and whether they are larger than 15mm in any direction (a dime is 17mm).

Neuroblastoma

Have you or a relative been diagnosed with neuroblastoma?

- ○ No / not that I am aware of
- ○ Yes
- ○ Other…

Psychiatric disease

Have you been diagnosed with any of the following psychiatric diseases? (click all that apply)

- ☐ Major depression
- ☐ Bipolar disorder
- ☐ Schizoaffective disorder
- ☐ Schizophrenia
- ☐ Other…

Benign neonatal seizures

Did you or a relative have benign seizures when an infant, during the first month of life, that went away?

- ○ No / not that I am aware of
- ○ Yes
- ○ Other…

Neuralgic amyotrophy

Neuralgic amyotrophy is a rare disease characterized by sudden onset of severe pain in shoulder or upper limbs, and subsequent muscle atrophy. Have you or a relative been diagnosed with neuralgic amyotrophy?

- ○ No / not that I am aware of
- ○ Yes
- ○ Other…

Retinitis pigmentosa

Autosomal dominant retinitis pigmentosa is characterized by progressive late onset vision loss, beginning with loss of night vision and peripheral vision. Do you or a relative have retinitis pigmentosa or similar symptoms?

- ○ No / not that I am aware of
- ○ Maybe, I or a relative has similar symptoms but has never been formally diagnosed
- ○ Yes, I or a relative has been diagnosed with retinitis pigmentosa
- ○ Other…

Hemolytic-uremic syndrome

Hemolytic-uremic syndrome is a severe acute episode requiring hospitalization and is often lethal. Have you or a relative had one or more episodes of hemolytic-uremic syndrome?

- ○ No / not that I am aware of
- ○ Yes
- ○ Other…

Thrombotic thrombocytopenic purpura

Yes

Other...

Don't touch!

We use this to associate your answers with your profile at my.personalgenomes.org.

Short answer text