| 2 3 | | | | | | | | | | | | |
|--|------------|-------------------------------|---|----------|---|---|----------------|--------------------------------|----------|---|-----|--|
| 4 5 | | | | | | | | | | | | |
| 6 7 | | P9: c.94C>T (p.P32S) | | | | | P7 | P7: c.395C>G (p.S132C) | | | | |
| 8 9 10 11 12 13 14 15 16 17 18 19 | variant | N | Κ | <u>s</u> | I | L | E | G | <u>C</u> | I | L | |
| | Human | N | Κ | Р | | L | E | G | S | 1 | L | |
| | Chimpanzee | N | κ | Р | 1 | L | E | G | S | 1 | L | |
| | Mouse | N | к | Р | 1 | L | E | G | S | 1 | L | |
| | Dog | N | к | Р | | L | E | G | S | I | L | |
| | Chicken | - | - | - | - | ÷ | E | G | S | 1 | V | |
| | Fruit fly | N | K | Р | 1 | L | E | G | Т | 1 | V | |
| | Roundworm | Ν | к | Р | М | М | Е | G | Т | 1 | A | |
| 20 21 | | | | | | | 2 | | | · | Ų. | |
| 22 | | | | | | | | | | | | |
| 23 24 | | P5: c.721C>T (p.P241S) | | | | | P5 | P5: c.1034T>C (p.V345A) | | | | |
| 25 26 27 28 29 30 31 32 33 34 35 36 | variant | K | Q | <u>S</u> | E | R | L | Е | A | R | М | |
| | Human | к | Q | Р | Е | R | L | Е | V | R | М | |
| | Chimpanzee | к | Q | Р | Е | R | 5 - | - | V | R | М | |
| | Mouse | к | Н | Р | Е | R | : . | : - | V | R | М | |
| | Dog | к | Q | Р | Е | Q | 7- | - | V | R | М | |
| | Chicken | Q | Н | Р | Е | К | :: | 19 1. 11 | V | R | М | |
| | Fruit fly | K | Q | S | Р | к | - <u>-</u> | (1 | V | R | l I | |
| | Roundworm | Т | к | S | D | к | | | С | R | L | |
| 37 38 | | | | | | | | | | | | |

Supp. Figure S1: Alignment of amino acids at sites of novel sequence variants.

Sites of novel sequence variants reported here in subjects P5, P7, and P9 are depicted in the alignments of various species from human to roundworm. Darker shading indicates a greater degree of amino acid conservation.