

# *add-1(syb1598)*

Strain name: PHX1598

Wild-type gene length: 1951bp

Mutation: 1539bp deletion at position 69 (second half of exon 1 to exon 5)

## *C. elegans* Description

Function: Encodes cytoskeletal alpha-adducin; associated with cell membrane-mediated localised changes in cytoskeleton in response to activated signal transduction cascades

Expression: Coelomocytes; neurons; intestinal and rectal epithelia

Previously reported phenotypes: Defective short/long term memory and defective GLR-1 ionotropic glutamate receptor dynamics<sup>1</sup>

## Human Orthologs:

*ADD1; ADD3*

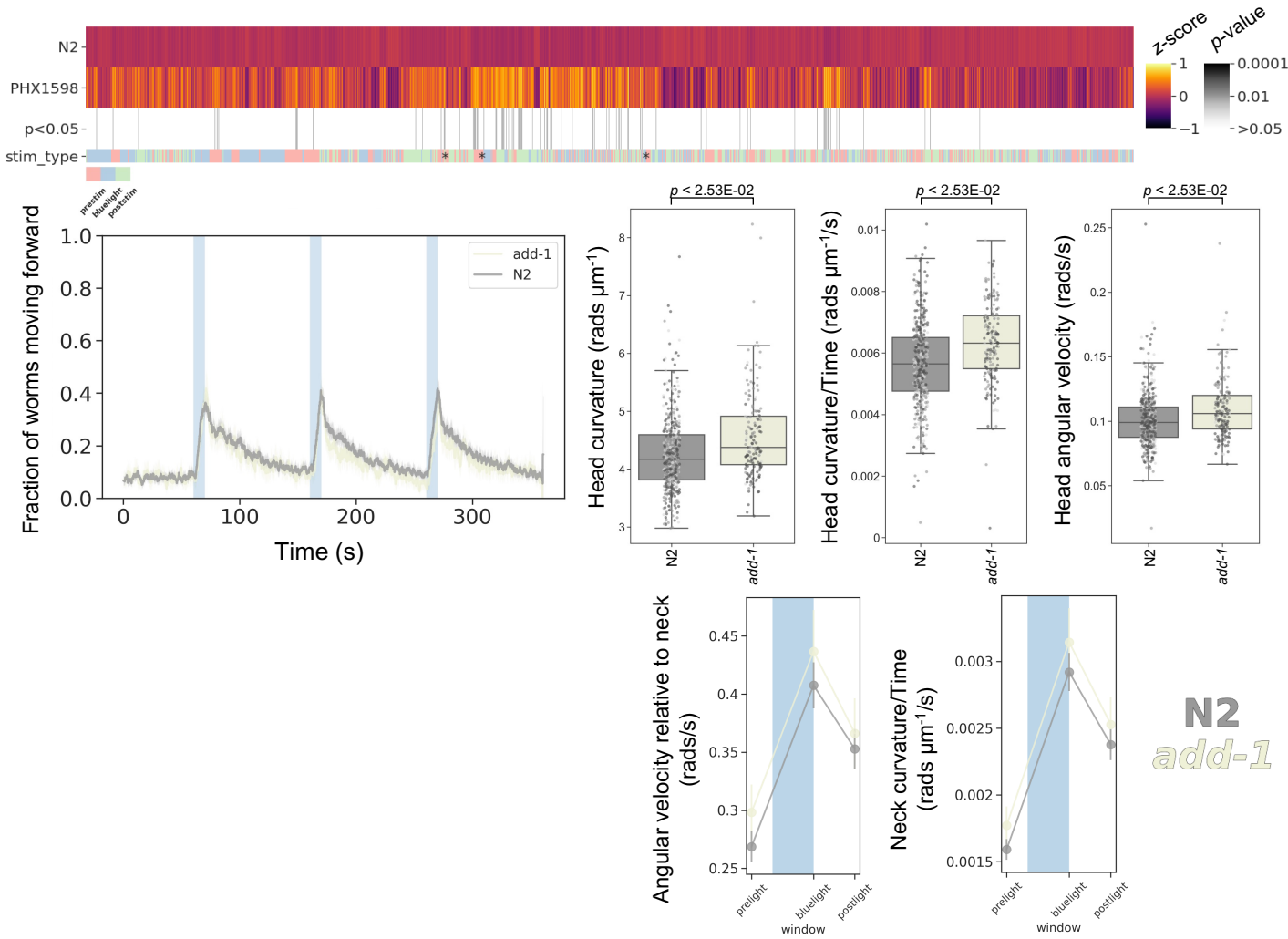
Associated disease(s):

Artery disease; cerebral palsy, spastic quadriplegic, 3, autosomal recessive (AR) 617008; Hypertension, essential, salt-sensitive, mutational (Mu) 145500

## Results

494/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): increased curvature and angular velocity of the head



# avr-14(syb1576)

Strain name: PHX1576

Wild-type gene length: 1111bp

Mutation: 628bp deletion at position 5 (exon 1 and start of exon 2)

## C. elegans Description

Function: Localises to membrane and encodes glutamate-gated chloride channel activity (ivermectin sensitive)

Expression: Neurons and somatic nervous system

Previously reported phenotypes: Defective salt-chemotaxis behaviour and decreased locomotion on food<sup>2-3</sup>

## Human Orthologs:

GLRA1, GLRB, GLRA3, GLRA4

## Associated disease(s):

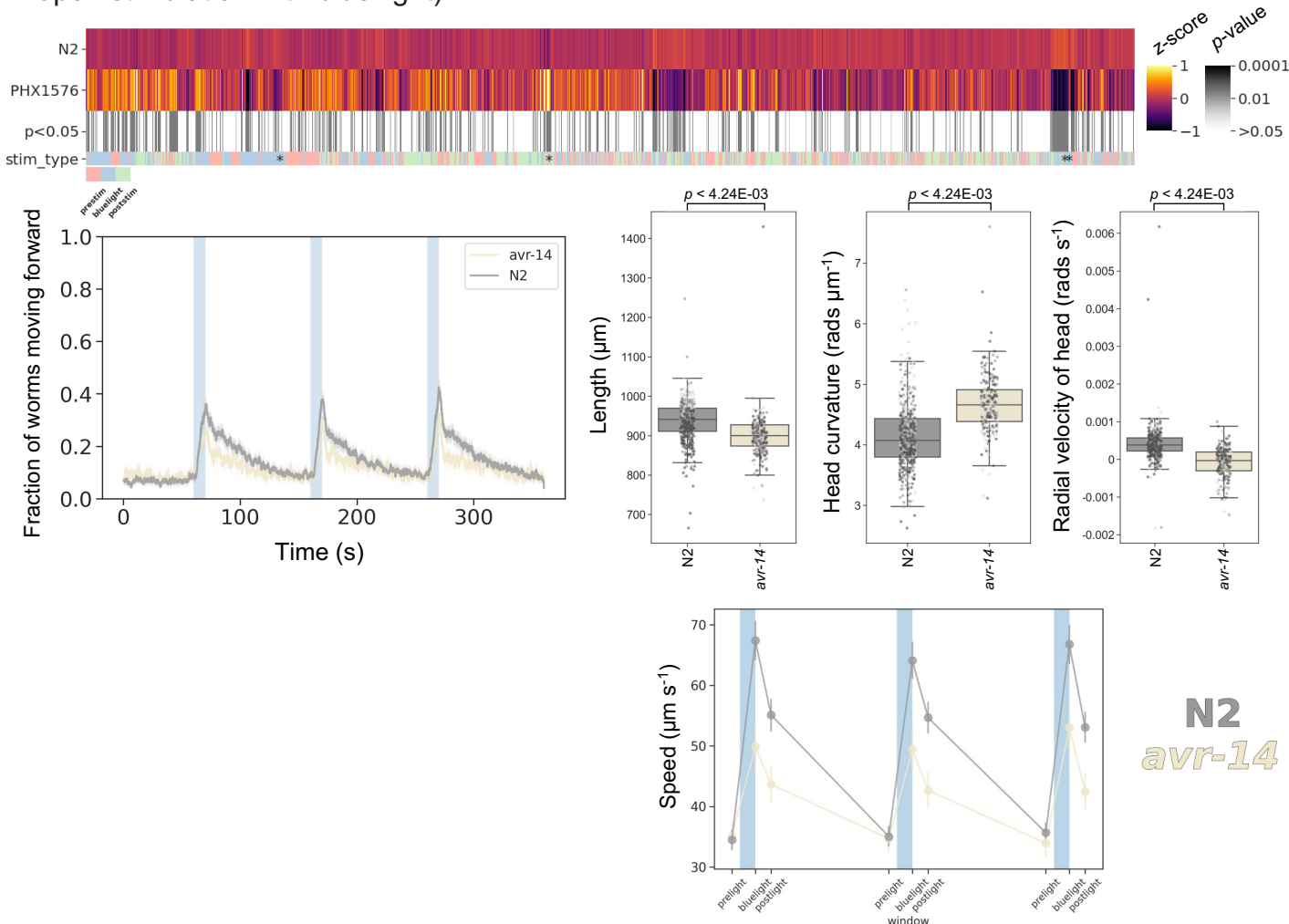
Hyperekplexia, hereditary 1, autosomal dominant (AD) or recessive (AR), 149400;

Hyperekplexia 2, 614619 (AR)

## Results

2717/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): small; increased curvature of the head and midbody; decreased head foraging (radial velocity); attenuated blue light response (e.g., decreased increase in speed upon stimulation with blue light)



# *bbs-1(syb1588)*

Strain name: PHX1588

Wild-type gene length: 1984bp

Mutation: 1412bp deletion at position 6 (exon 1 to start of exon 2)

## *C. elegans* Description

**Function:** Involved in intraciliary retrograde transport, non-motile cilium assembly and protein localization to cilium; regulates fat storage, as such, *bbs-1* affects body size, feeding, chemosensation and is required for proper regulation of insulin secretion

**Expression:** Ciliated neurons and sensory neurons

**Previously reported phenotypes:** Increased secretion of insulin, biogenic amines and neuropeptides; abnormal dye filling; ivermectin resistance<sup>4-5</sup>

## Human Orthologs:

*BBS1*

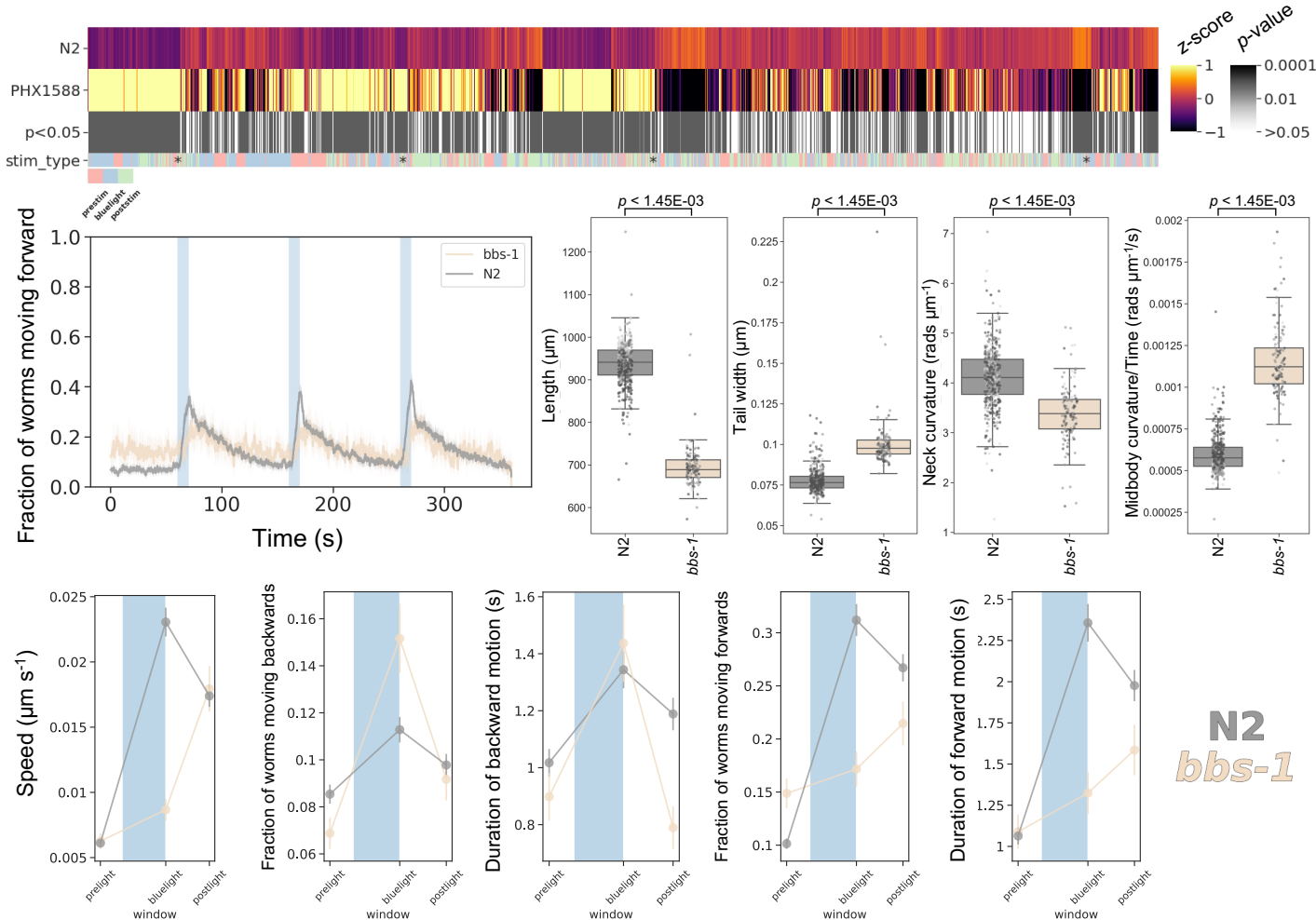
**Associated disease(s):**

Bardet-Biedl syndrome 1, autosomal recessive (AR) or digenic recessive (DR), 209900

## Results

6124/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): shorter and fatter; decreased curvature; deeper body bends (time derivative of curvature); hyperactive prior to simulation; delayed, but sustained, forward escape response and an increased, but short-lived, backward escape response to blue light



# *bbs-2(syb1547)*

Strain name: PHX1547

Wild-type gene length: 2330bp

Mutation: 1513bp deletion at position 91 (second half exon 1 to half of exon 4)

## *C. elegans* Description

Function: Involved in non-motile cillum assembly

Expression: Ciliated neurons and sensory neurons

Previously reported phenotypes: Body size, developmental delay and roaming defects<sup>4</sup>

## Human Orthologs:

*BBS2*

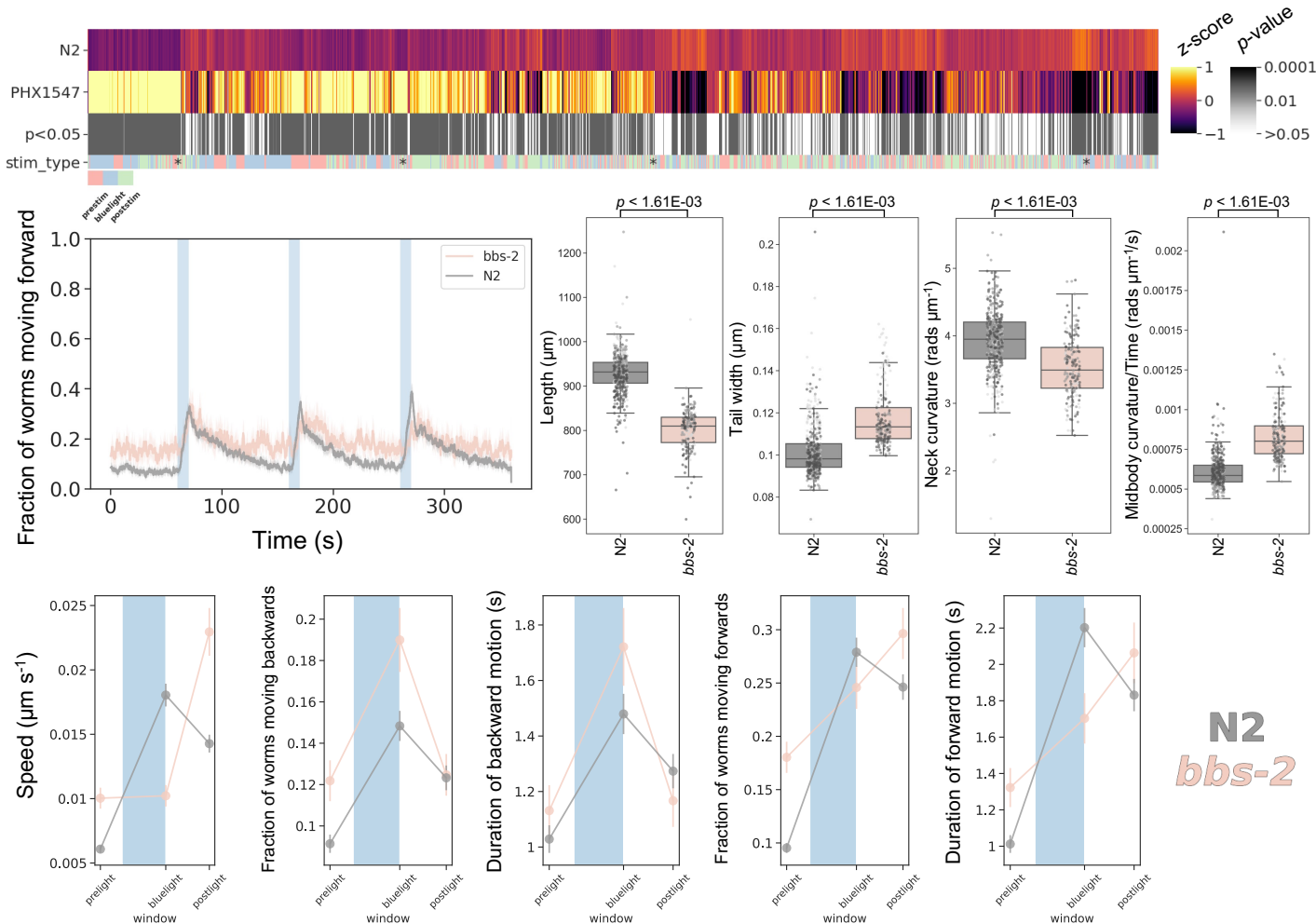
Associated disease(s):

Bardet-Biedl syndrome 2, 615981 (AR); Retinitis pigmentosa 74, 616562 (AR)

## Results

5693/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): shorter and fatter; decreased curvature; deeper body bends (time derivative of curvature); hyperactive prior to stimulation; increased (but delayed) increase in speed, short lived backward escape response, and delayed (but sustained) forward escape response to blue light stimulation





# cat-2(syb1542)

Strain name: PHX1542

Wild-type gene length: 1683bp

Mutation: 1520bp deletion at position 33 (half of exon 1 to half of exon 3)

## C. elegans Description

Function: Tyrosine hydroxylase activity (catecholamine synthesis)

Expression: Dopaminergic neurons and male-specific anatomy (spiracles and ray neurons)

Previously reported phenotypes: Decreased dopamine levels; defective food sensing; increased peak acceleration; increased variation in speed; increased habituation in response to nonlocalized mechanical stimulation<sup>6-7</sup>

## Human Orthologs:

TH

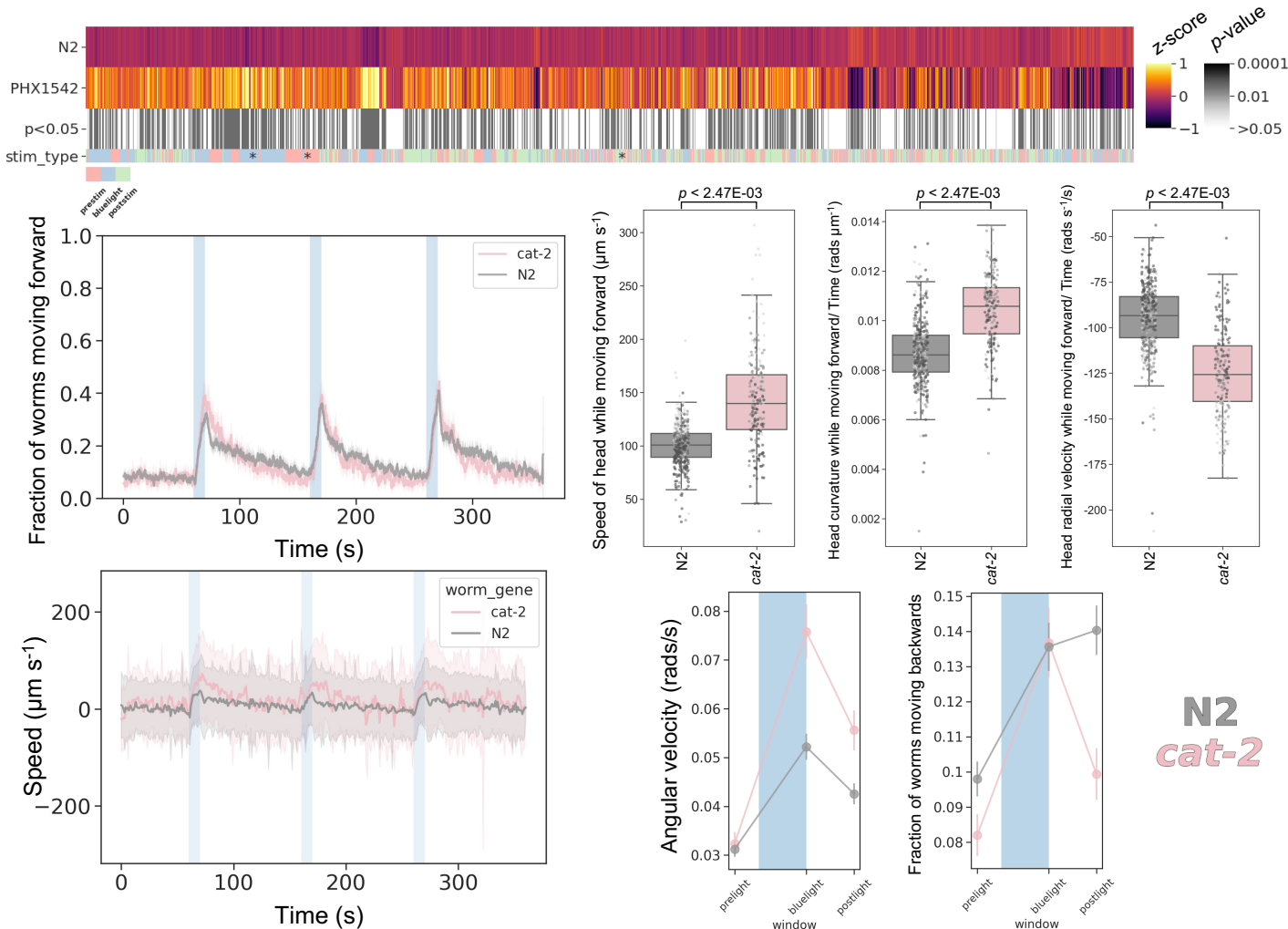
Associated disease(s):

L-Dopa-responsive dystonia/Segawa syndrome, 605407 (AR); Obesity; Parkinsonism

## Results

4479/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): increased speed/curvature of head tip and decreased head foraging (radial velocity of head) while moving forward; hypersensitive to blue light stimulation (e.g., greater increase in speed and angular velocity); short backward escape response to blue light



# cat-4(syb1591)

Strain name: PHX1591

Wild-type gene length: 1647bp

Mutation: 891bp deletion at position 384 (exons 2-3 and half of exon 4)

## C. elegans Description

Function: Putative GTP cyclohydrolase I activity

Expression: Muscle cells; pharynx motor neurons; intestinal cells; dopaminergic, serotonergic, and mechanosensory neurons

Previously reported phenotypes: No previous phenotype reported

## Human Orthologs:

*GHC1*

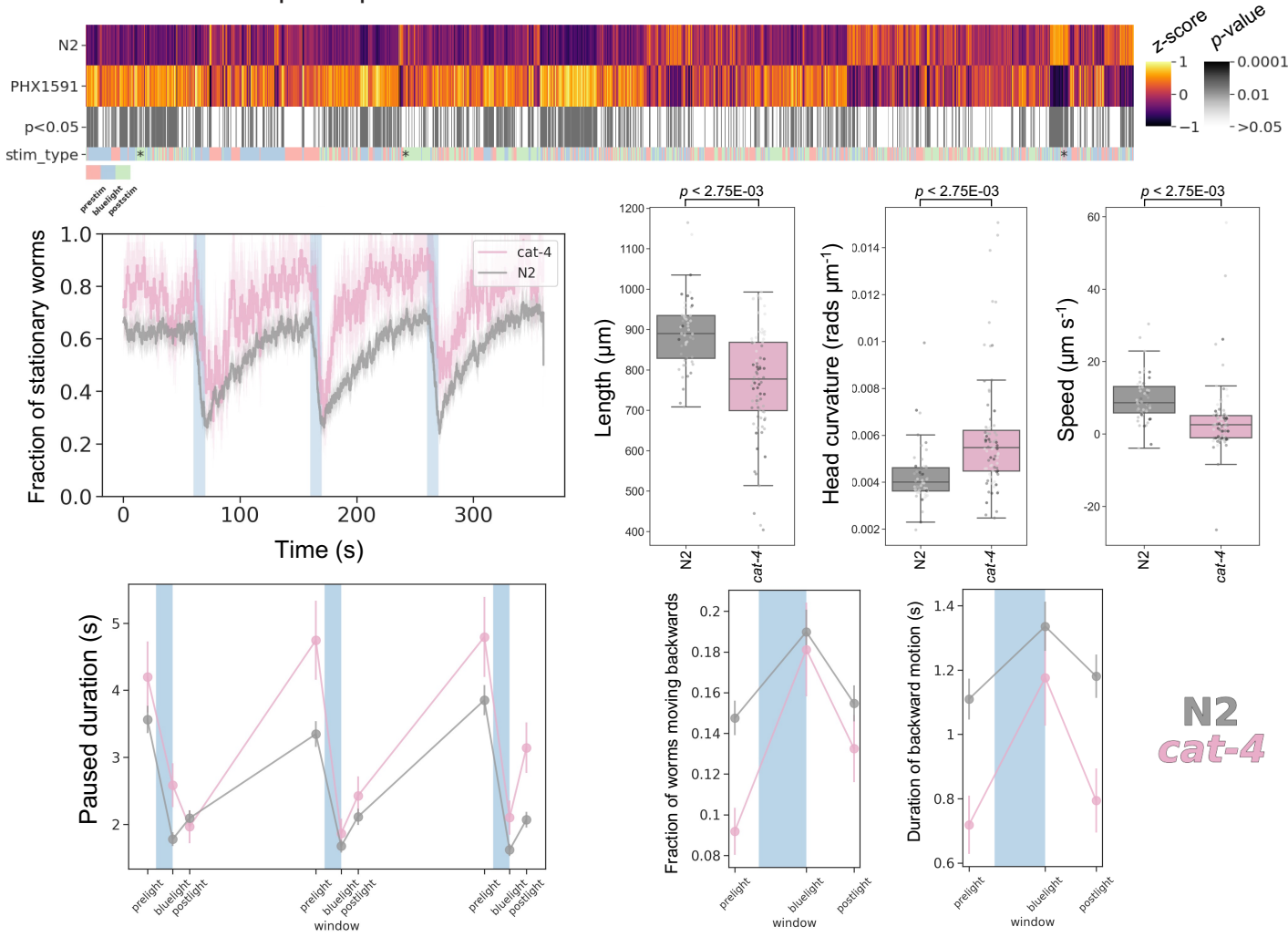
Associated diseases (from OMIM):

*L*-Dopa-responsive dystonia, with or without hyperphenylalaninemia, 128230 (AD, AR);  
Hyperphenylalaninemia, BH4 deficient, B, 233910 (AR); Bipolar disorder

## Results

4182/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): Developmental delay (2 days); small brood size; smaller; increased curvature; decreased speed; less active; strong photophobic escape response; short duration of backwards escape response



# *dys-1(syb1688)*

Strain name: PHX1688

Wild-type gene length: 30990bp

Mutation: 30716bp deletion at position 59 (all of the F32B4.10 transcript)

## *C. elegans* Description

Function: Localizes to dystrobrevin complex

Expression: Body wall musculature; head muscle; pharyngeal muscle; vulval muscle

Previously reported phenotypes: Subtle hyperactive locomotion; exaggerated head bending; slight hypercontractions; abnormal burrowing<sup>8-9</sup>

## Human Orthologs:

*DMD*

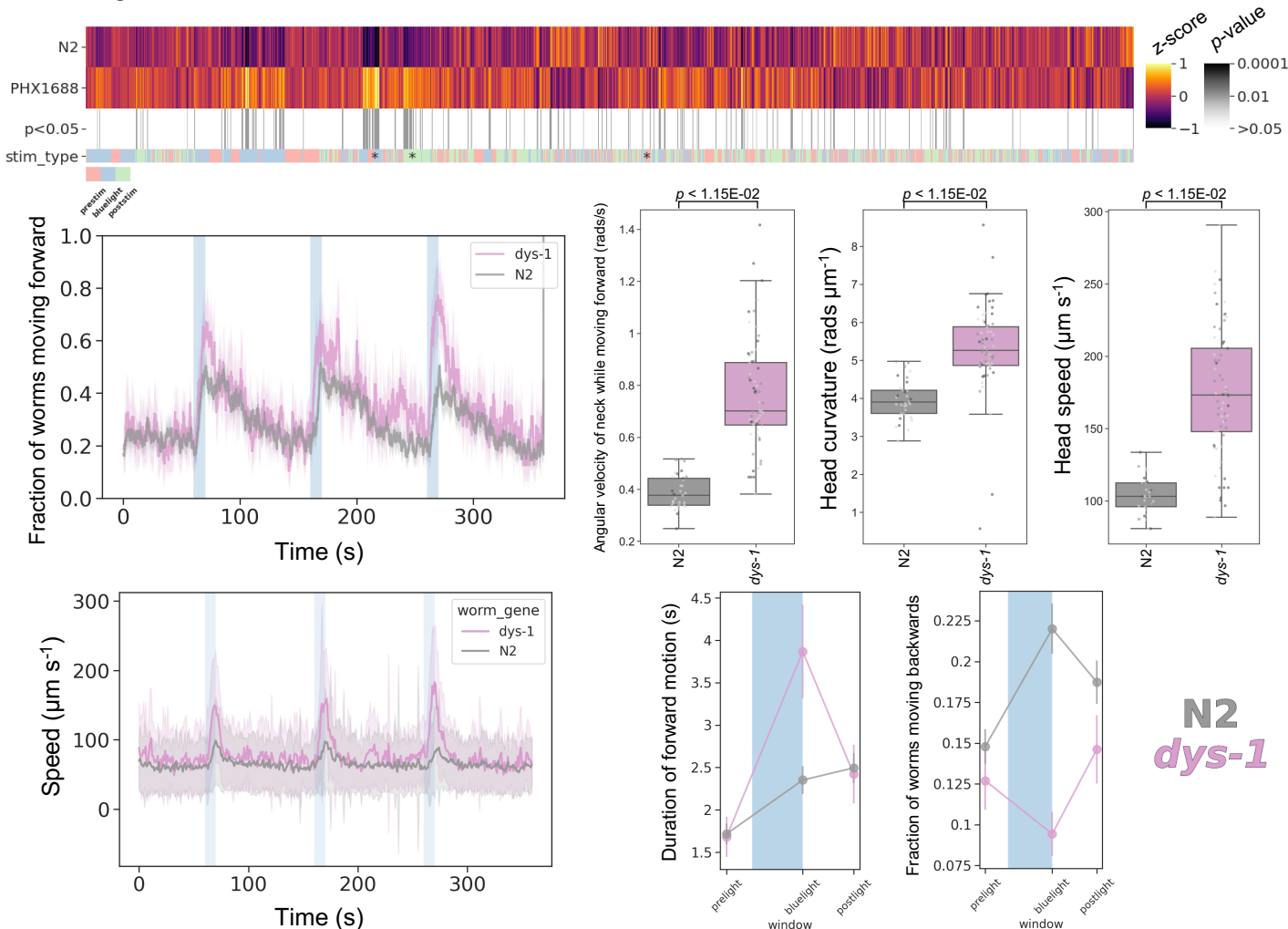
Associated disease(s):

Duchenne muscular dystrophy, X-linked recessive (XLR) 310200; Becker muscular dystrophy, 300376 (XLR); Cardiomyopathy dilated, 302045 (XLR)

## Results

1038/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): Developmental delay (4 days); increased curvature, angular velocity and speed of head; increased sensitivity to blue light stimuli (e.g., greater increase in speed/forward motion upon stimulation); strongly attenuated backward escape response to blue light stimulation



# figo-1(syb1562)

Strain name: PHX1562

Wild-type gene length: 1633bp

Mutation: 1038bp deletion at position 18 (removing the first 3 exons)

## C. elegans Description

Function: Putative phosphoric ester hydrolase activity

Expression: GABAergic neurons; body wall musculature; head neurons; intestine and tail neurons

Previously reported phenotypes: No previous phenotype reported

## Human Orthologs:

*FIG4*

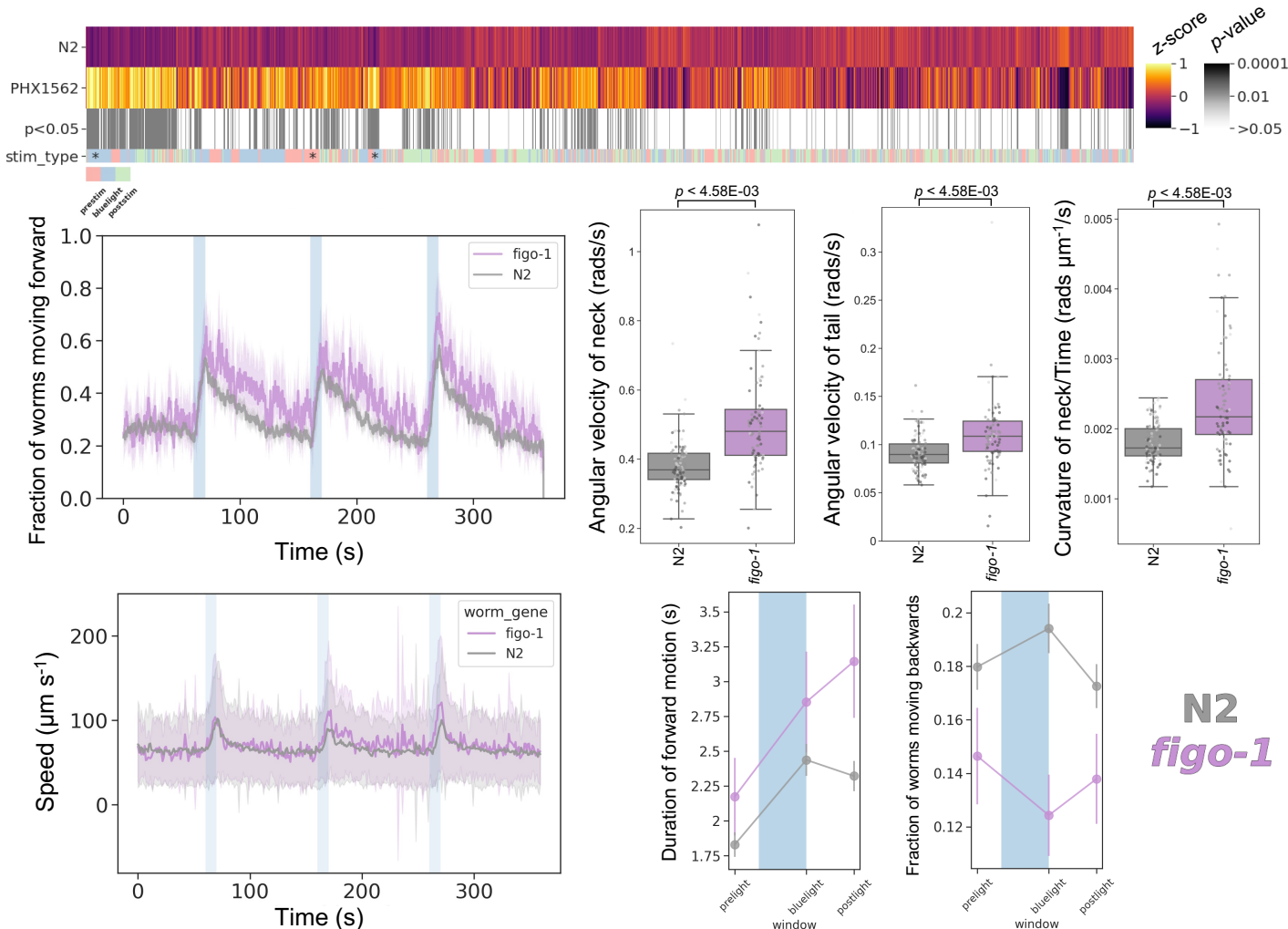
Associated disease(s):

Amotrophic lateral sclerosis 11, 612577 (AD); Charcot-Marie-Tooth disease, type 4J, 611228 (AR); Polymicrogyria, bilateral temporooccipital, 612691 (AR); Yunis-Varon syndrome, 216340 (AR)

## Results

2799/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): Developmental delay (2 days); increased angular velocity/curvature of the head, neck and tail; deeper body bends (time derivative of curvature); sustained forward, but attenuated backward, escape response to blue light stimulation; greater increase in speed upon repeated blue light stimulation



# glc-2(syb1557)

Strain name: PHX1557

Wild-type gene length: 1512bp

Mutation: 1269bp deletion at position 106 (first 4 exons)

## C. elegans Description

**Function:** Glutamate-gated Chloride channel (GLC-2), capable of forming homomeric glutamate-activated channels and heteromeric glutamate-activated channels with GLC-1; avermectins or anthelmintics inhibit pharyngeal pumping

**Expression:** Pharyngeal pm4 muscle

**Previously reported phenotypes:** No previous phenotype reported

## Human Orthologs:

GLRA1; GLRB

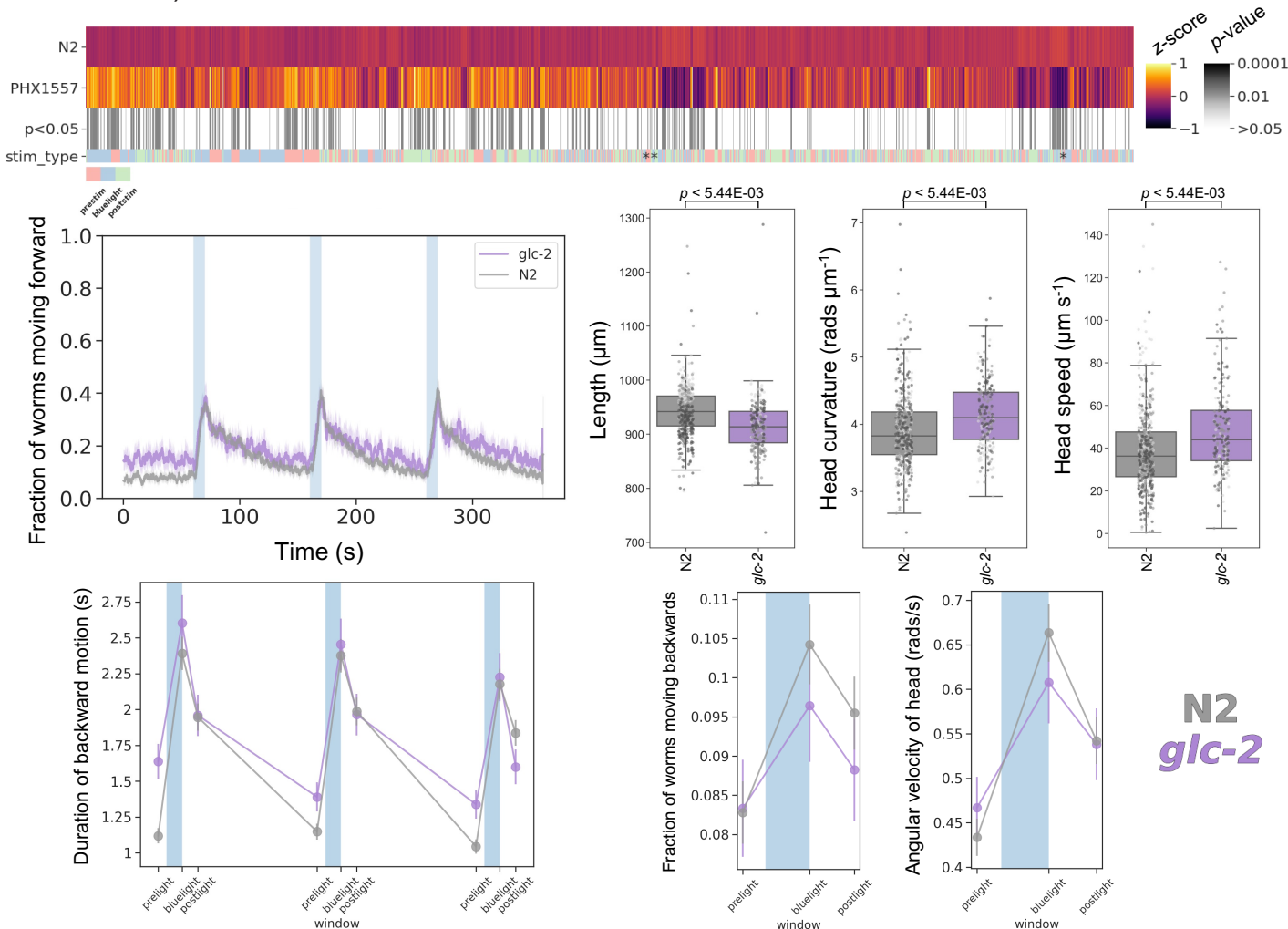
## Associated disease(s):

Hyperekplexia, hereditary 1, 149400 (AD or AR); Hyperekplexia 2, 614619 (AR)

## Results

2413/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

**Key phenotype(s):** Smaller; increased curvature and speed of the head; hyperactive; increased fraction and duration of forward motion; attenuated backward escape response; diminished blue light response (e.g., change in angular velocity upon stimulation is decreased)



# *glr-1(syb1556)*

Strain name: PHX1556

Wild-type gene length: 1488bp

Mutation: 979bp deletion at position 10 (exon 1 to beginning of exon 4)

## *C. elegans* Description

Function: Localizes to several cellular components, including: ionotropic glutamate receptor complex; neuron projection membrane and perinuclear endoplasmic reticulum

Expression: Ganglia; neurons and ventral nerve cord

Previously reported phenotypes: Gain of function mutants are resistant to dopamine-induced food paralysis and have higher reversal rate; loss of function mutants are defective in light nose touch response; resistance to fluoxetine-induced paralysis<sup>10-12</sup>

## Human Orthologs:

*GRIA3, GRIK2*

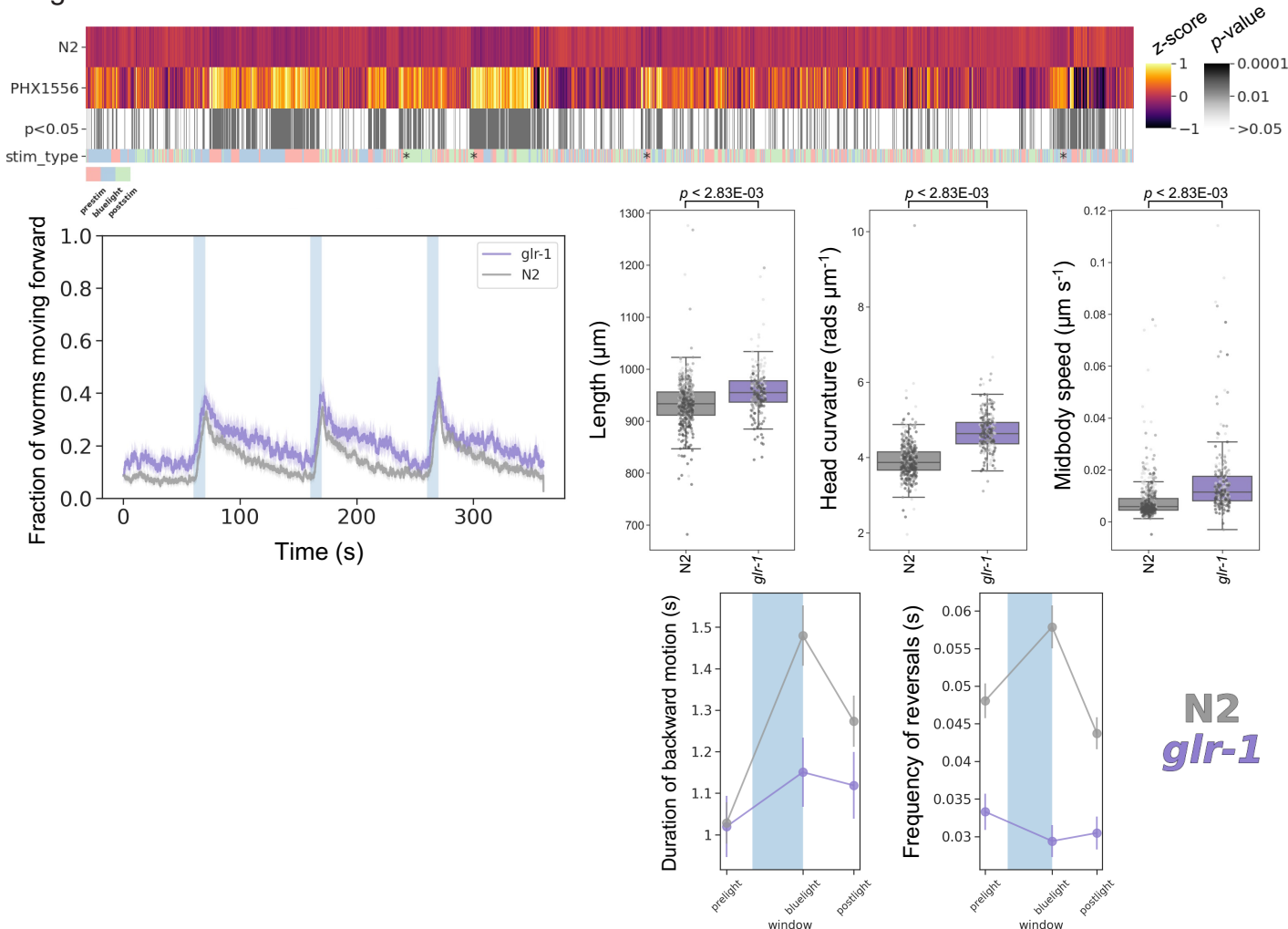
Associated disease(s):

Intellectual disability, 94, 300699 (XLR); Intellectual disability, 6, 611092 (AR); Huntington's

## Results

3820/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): Longer; increased foraging; increased curvature/angular velocity of all body segments; increased speed; hyperactive; attenuated backward escape response to blue light





# *glr-4(syb1518)*

Strain name: PHX1518

Wild-type gene length: 2124bp

Mutation: 1356bp deletion at position 103 (end of exon 1 to exon 8)

## *C. elegans* Description

Function: NMDA-receptor subunit (encodes a putative non-NMDA ionotropic glutamate receptor subunit)

Expression: DA9; SABVL; SABVR and VB neurons

Previously reported phenotypes: Aldicarb resistance<sup>13</sup>

## Human Orthologs:

*GRIK2*

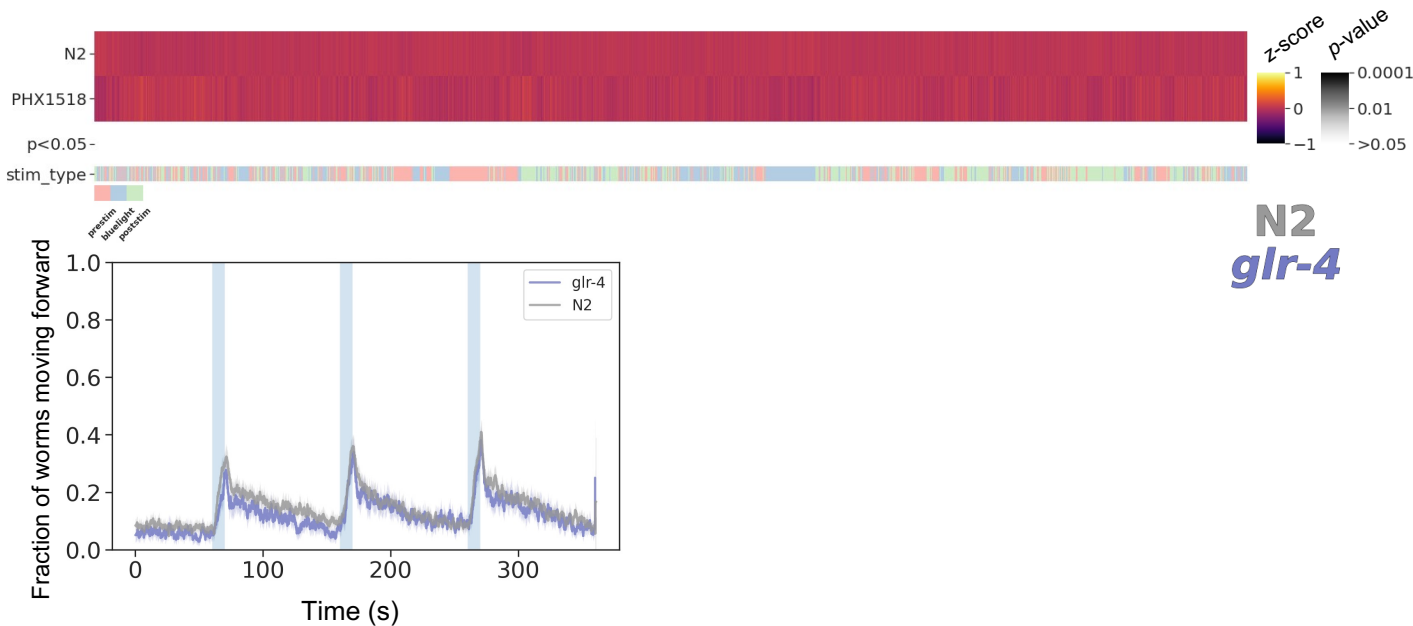
Associated disease(s):

Intellectual disability, 6, 611092 (AR)

## Results

0/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): No significant phenotype detected





# *gpb-2(syb1577)*

Strain name: PHX1577

Wild-type gene length: 3009bp

Mutation: 2408bp deletion at position 140 (all exons)

## *C. elegans* Description

Function: G protein-coupled acetylcholine receptor signalling pathway; egg-laying

Expression: Body wall musculature; nerve ring; pharyngeal muscle cell and vulval muscle

Previously reported phenotypes: Slippery corpus; slightly longer; increased sinusoidal and deeper body bends; defects in egg laying and pharyngeal pumping; arecoline sensitive<sup>14-16</sup>

## Human Orthologs:

*GNB5*

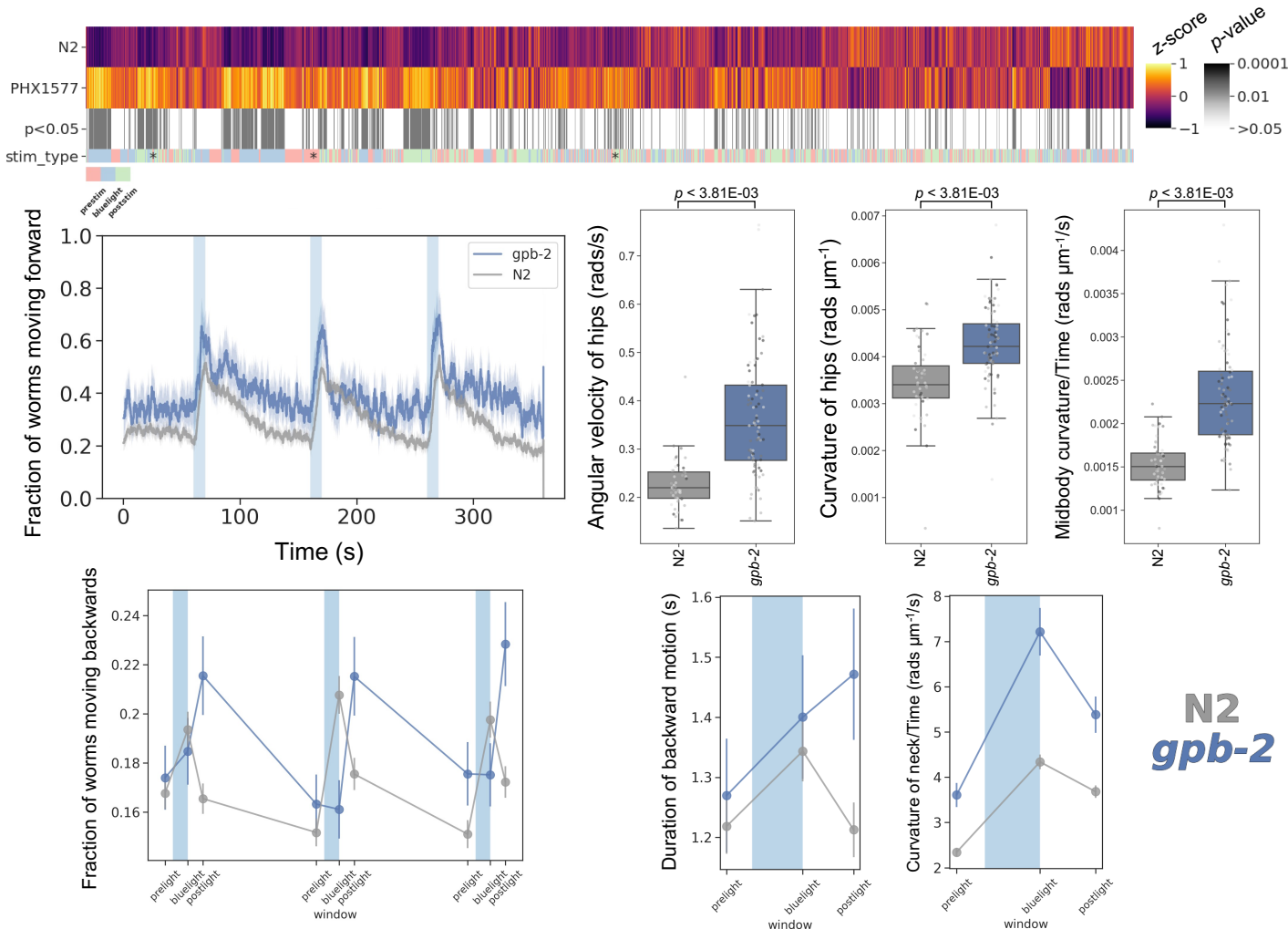
### Associated disease(s):

Intellectual developmental disorder with cardiac arrhythmia, 617173 (AR); Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182 (AR)

## Results

2954/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): Developmental delay (2 days); increased curvature/angular velocity of hips and tail; increased sinusoidal movements (time derivative of curvature); hyperactive; stronger, more sustained, backward escape response; hypersensitive to blue light



# *kcc-2(syb2673)*

Strain name: PHX2673

Wild-type gene length: 4387bp

Mutation: 3919bp deletion at position 45 (first 3 exons and most piwi-interacting RNAs)

## *C. elegans* Description

**Function:** Potassium-chloride c-transporter (partially redundant with *abts-1*); involved in chloride/potassium ion transport and regulation of GABAergic synaptic transmission

**Expression:** Body wall musculature; egg-laying apparatus; hermaphrodite distal tip cell; intestine and neurons

**Previously reported phenotypes:** Decreased egg laying; shorter; serotonin resistant; displays an excitatory, not inhibitory, response to the GABAa agonist muscimol<sup>17-18</sup>

## Human Orthologs:

*SLC12A5, SLC12A6*

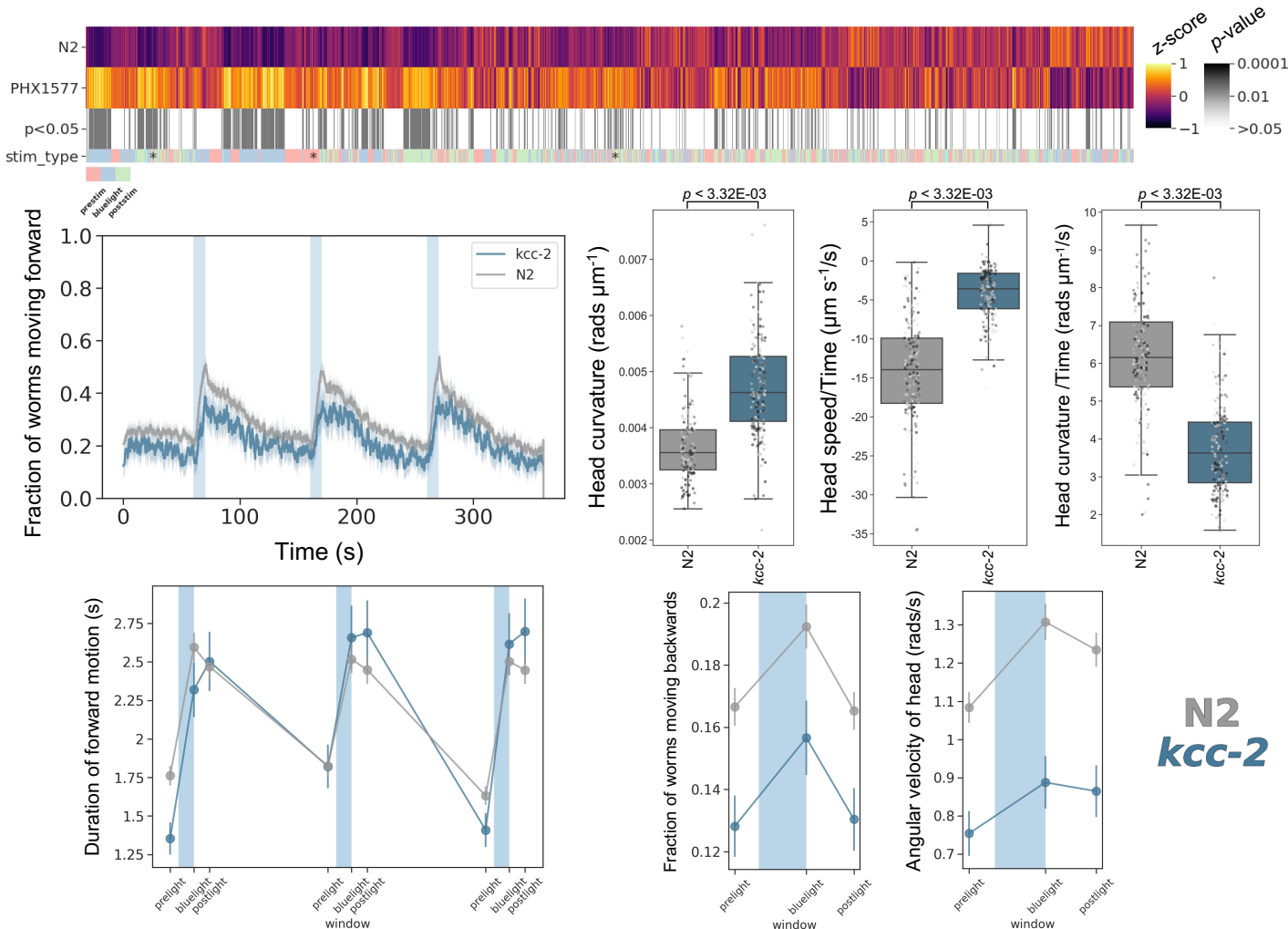
## Associated disease(s):

Epilepsy, idiopathic generalized, 14, 616685 (AD); Epileptic encephalopathy, early infantile, 34, 616645 (AR); Agenesis of the corpus callosum with peripheral neuropathy, 218000 (AR)

## Results

3246/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): Developmental delay (2 days); increased curvature and speed of head; decreased sinusoidal undulations (time derivative of midbody curvature); decreased angular velocity; hypoactive; sustained forward escape response; attenuated blue light response



# mpz-1(syb1522)

Strain name: PHX1522

Wild-type gene length: 36850bp

Mutation: 35787bp deletion at position 423 (entire gene)

## C. elegans Description

Function: Encodes multi-PDZ domain scaffold protein that exhibits GTPase activating protein binding activity

Expression: Body wall musculature; nerve ring; neurons and vulval muscle

Previously reported phenotypes: *mpz-1* RNAi reduces 5-HT stimulated egg laying<sup>19</sup>

## Human Orthologs:

*MPDZ*

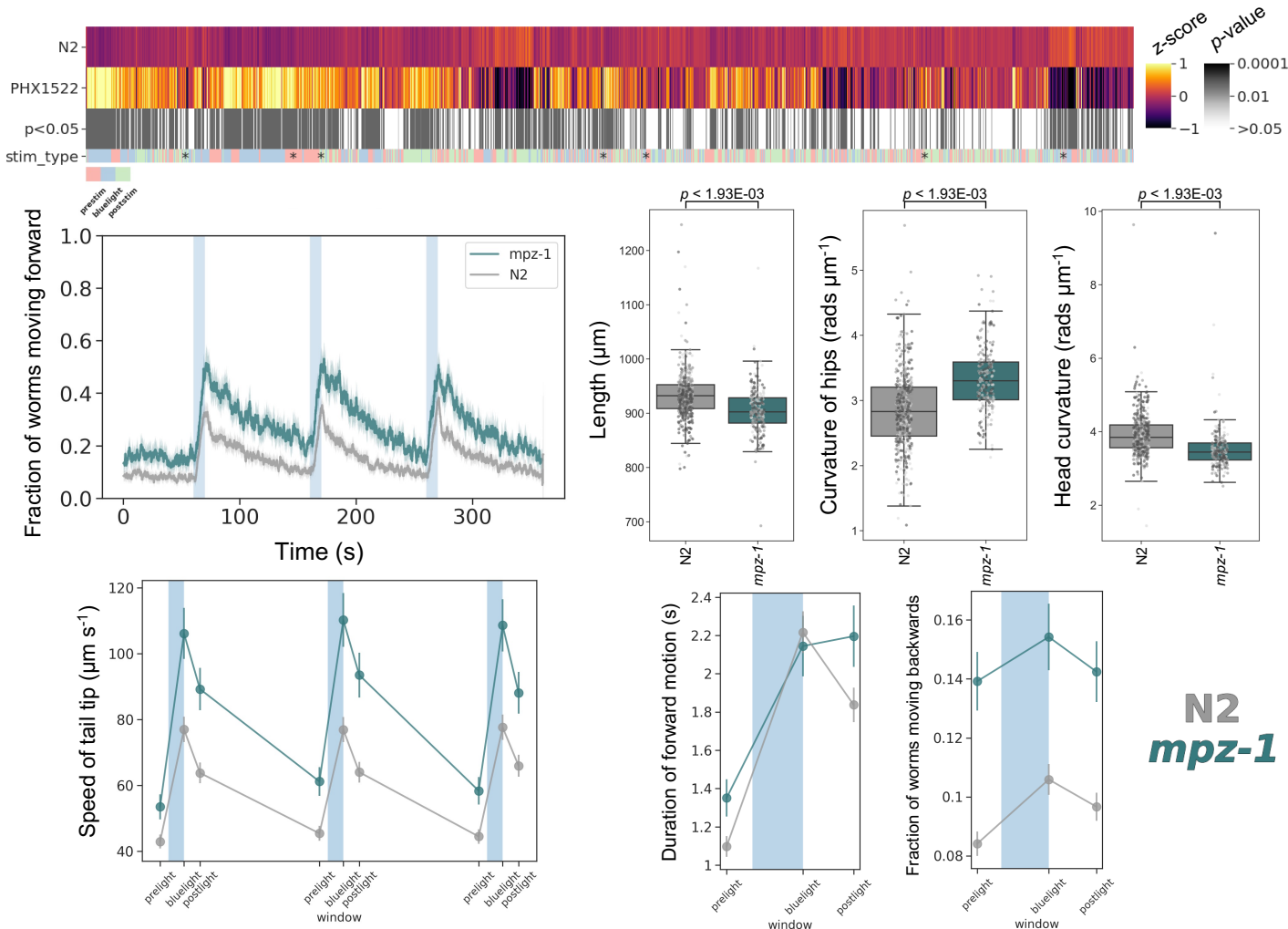
Associated disease(s) :

Hydrocephalus, nonsyndromic, 2, 615219 (AR)

## Results

4957/8289 significant features vs ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): Shorter; decreased curvature of the head/neck but increased curvature of hips/midbody; increased speed (hypersensitive to blue light); hyperactive; sustained forward escape response; increased rate of reversals (fraction of backwards moving worms)



# *nca-2(syb1612)*

Strain name: PHX1612

Wild-type gene length: 8377bp

Mutation: 6003bp deletion at position 20 (first 13.5 exons)

## *C. elegans* Description

Function: Cation leak channel activity; interacts with *unc-77* and *unc-80*

Expression: Intestine; pharyngeal neurons and somatic nervous system

Previously reported phenotypes: Locomotion deficits; fainter in liquid; sensitive to halothane and isoflurane<sup>15</sup>

## Human Orthologs:

*NALCN*

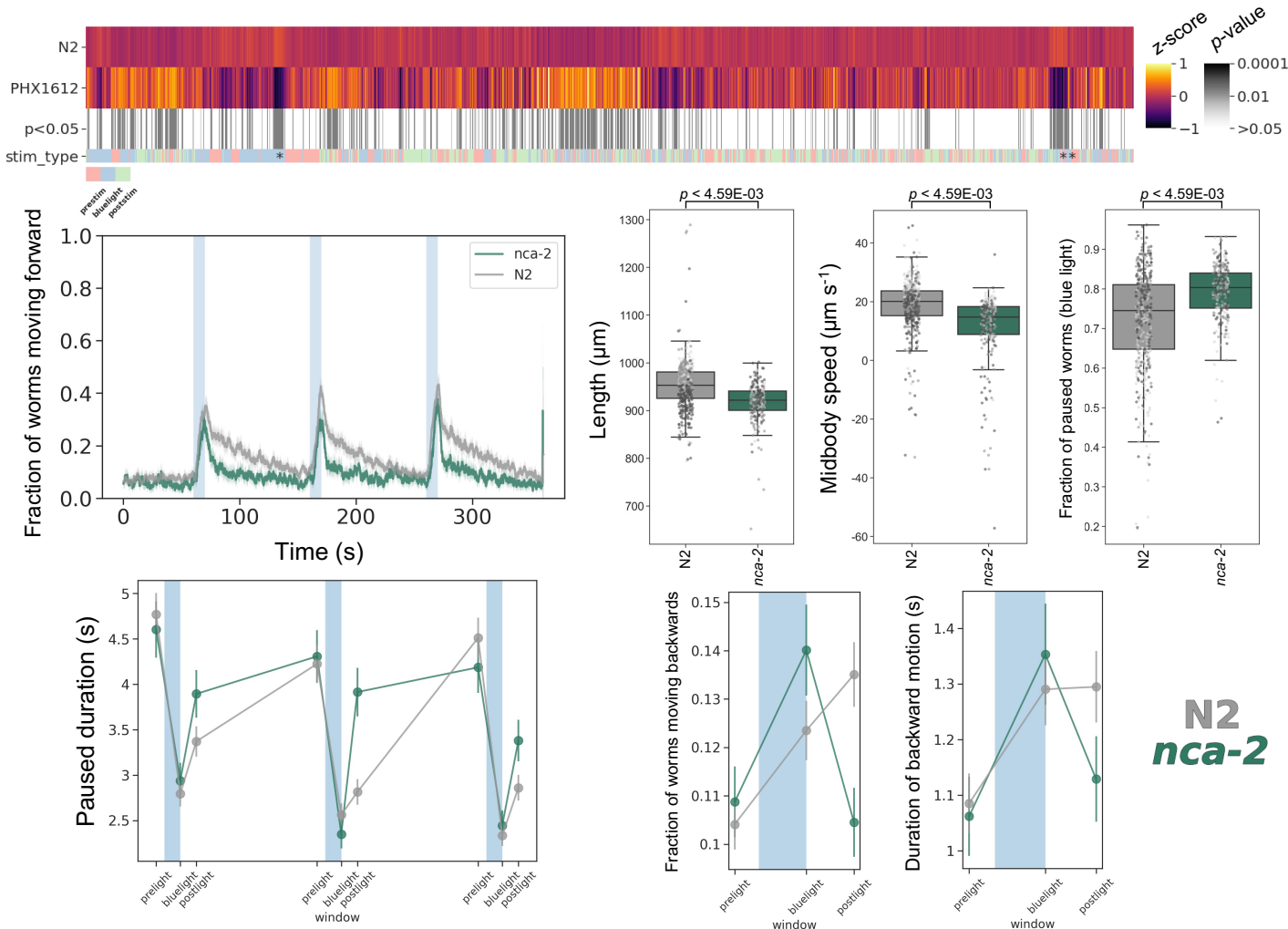
### Associated disease(s):

Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 (AD); Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 (AR)

## Results

2522/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): Smaller; decreased speed; increased curvature/angular velocity of head only; no difference in baseline locomotion but an increased, short-lived, reversal response to blue light; increased post-stimulation pausing, "fainting" phenotype



# *pink-1(syb1546)*

Strain name: PHX1546

Wild-type gene length: 1926bp

Mutation: 1381bp deletion at position 157 (second half exon 2 to exon 6)

## *C. elegans* Description

Function: Serine/threonine-specific protein kinase

Expression: Hermaphrodite distal tip cell; intestine; neurons; pharynx and vulva

Previously reported phenotypes: Required for: normal response to oxidative stress; mitochondrial homeostasis; correct CAN neurite outgrowth and wild type brood sizes. Mutant phenotypes are suppressed by mutation in *lrk-1* (LRRK2 ortholog)<sup>20-22</sup>

## Human Orthologs:

*PINK1*

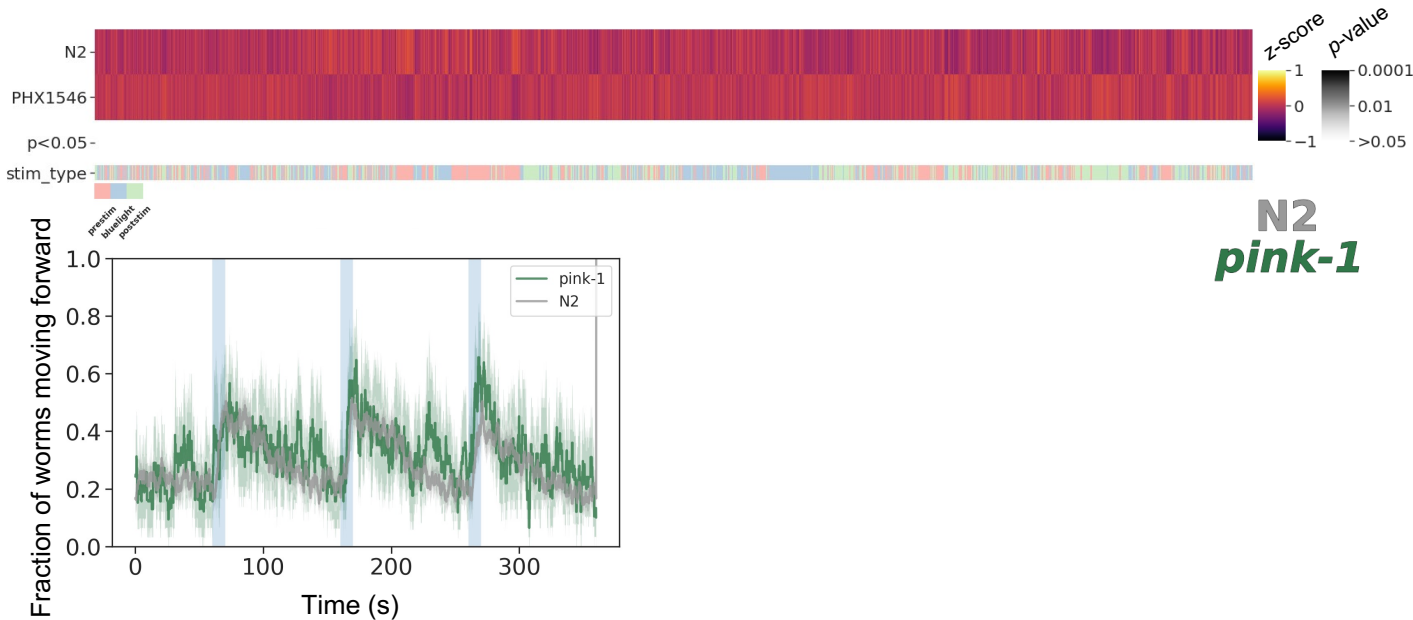
Associated disease(s):

Parkinson disease 6, early onset, 605909 (AR)

## Results

0/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): No significant phenotype detected



N2  
*pink-1*

# *snf-11(syb2683)*

Strain name: PHX2683

Wild-type gene length: 2283bp

Mutation: 1543bp deletion at position 16 (exon 1 to second half of exon 4)

## *C. elegans* Description

Function: Sodium/chloride ion-coupled high affinity GABA transporter; involved in gamma-aminobutyric acid transport

Expression: GLR; enteric muscle; head mesodermal cell; neurons and ventral nerve cord

Previously reported phenotypes: Abnormal head foraging and sinusoidal movements<sup>15</sup>

## Human Orthologs:

*SLC6A1*

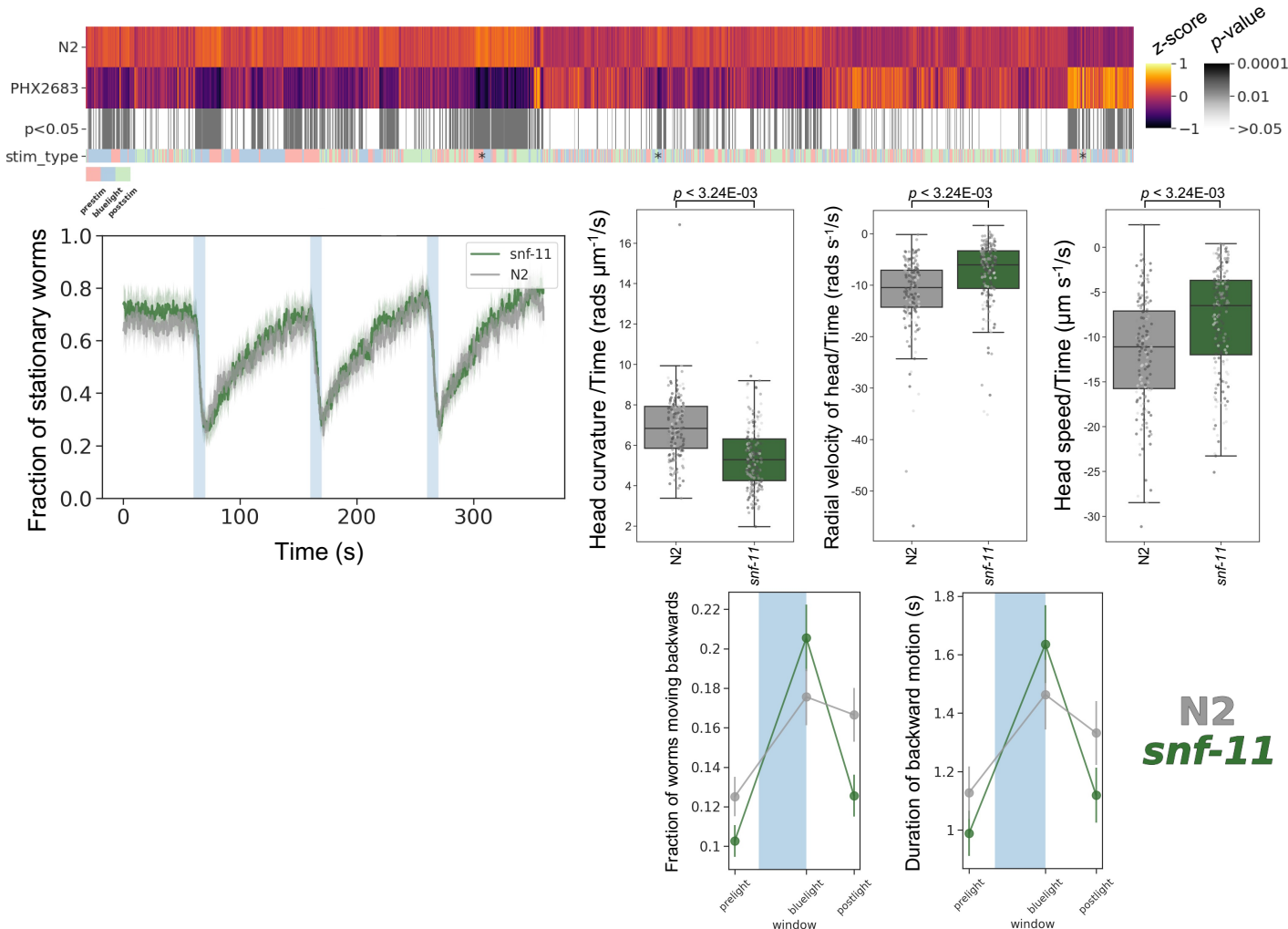
Associated disease(s):

Myoclonic-atonic epilepsy, 616421 (AD)

## Results

3601/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): slightly less active; low frequency head bends (decreased time derivative of curvature); increased speed of head; increased foraging behaviour (time derivative of radial velocity); strong, but short-lived, backwards escape response to blue light stimulation





# *snn-1(syb2695)*

Strain name: PHX2695

Wild-type gene length: 2682bp

Mutation: 1835bp deletion at position 22 (2 exons)

## *C. elegans* Description

Function: Synapsin, involved in neuromuscular synaptic transmission

Expression: All neurons; localises to presynaptic regions

Previously reported phenotypes: Aldicarb resistance; defective synaptic vesicle clustering<sup>13,23</sup>

## Human Orthologs:

*SYN1;SYN2*

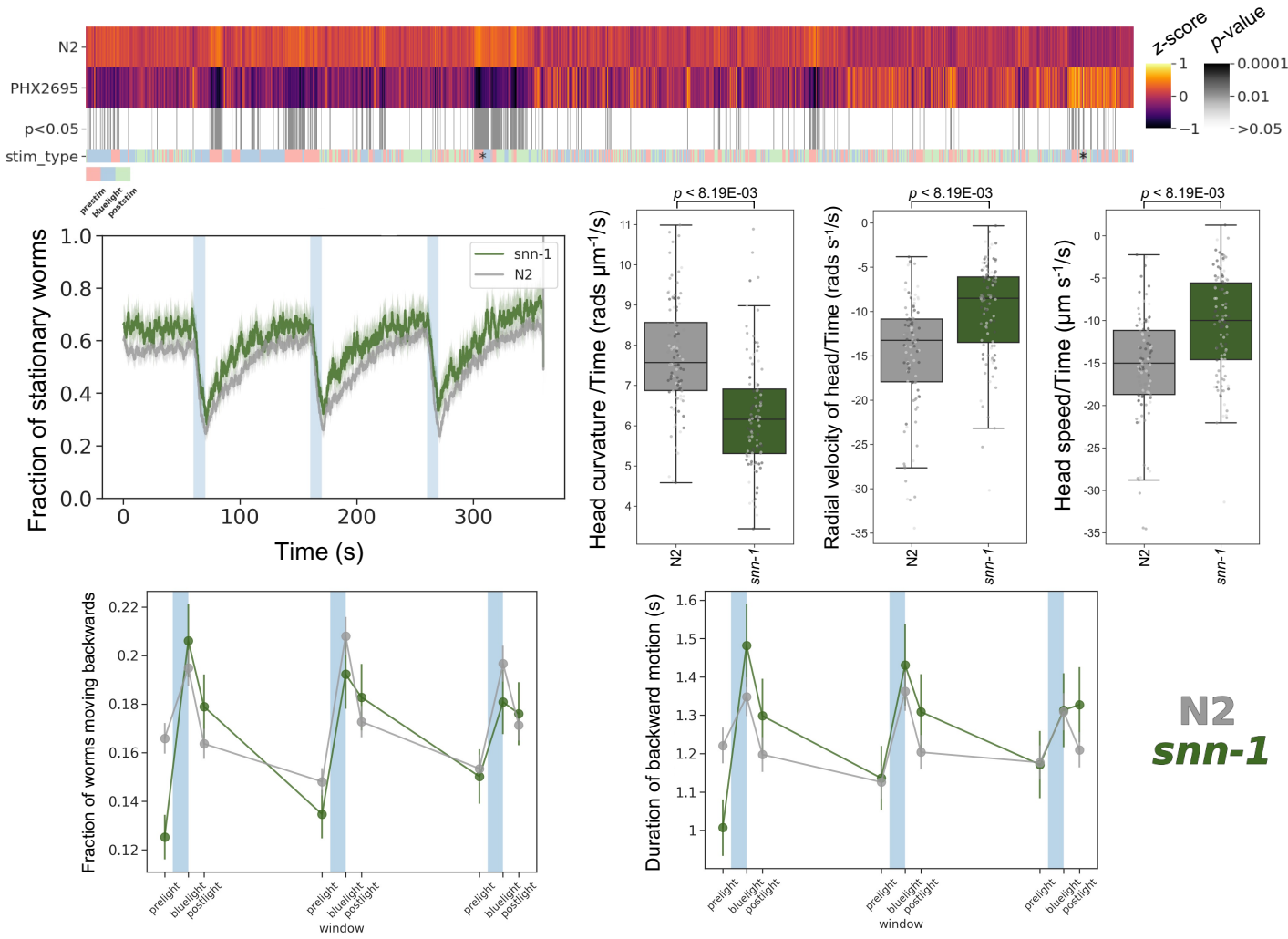
Associated disease(s):

Epilepsy, X-linked recessive and dominant (XLR, XLD), with variable learning disabilities and behaviour disorders, 300491; Schizophrenia susceptibility, 181500 (AD)

## Results

1633/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): Less active; shallow head movements (decreased time derivative of curvature); increased head speed; increased foraging behaviour (time derivative of radial velocity); increased reversal response to blue light that diminishes upon repeated exposure





# *tmem-231(syb1575)*

Strain name: PHX1575

Wild-type gene length: 2033bp

Mutation: 1253bp deletion at position 354 (covering exon 1 to beginning of exon 7)

## *C. elegans* Description

Function: Involved in non-motile cilium assembly

Expression: Amphid neurons; ciliated neurons; inner labial neurons; outer labial neurons and phasmid neurons

Previously reported phenotypes: No previous phenotype reported

## Human Orthologs:

*TMEM231*

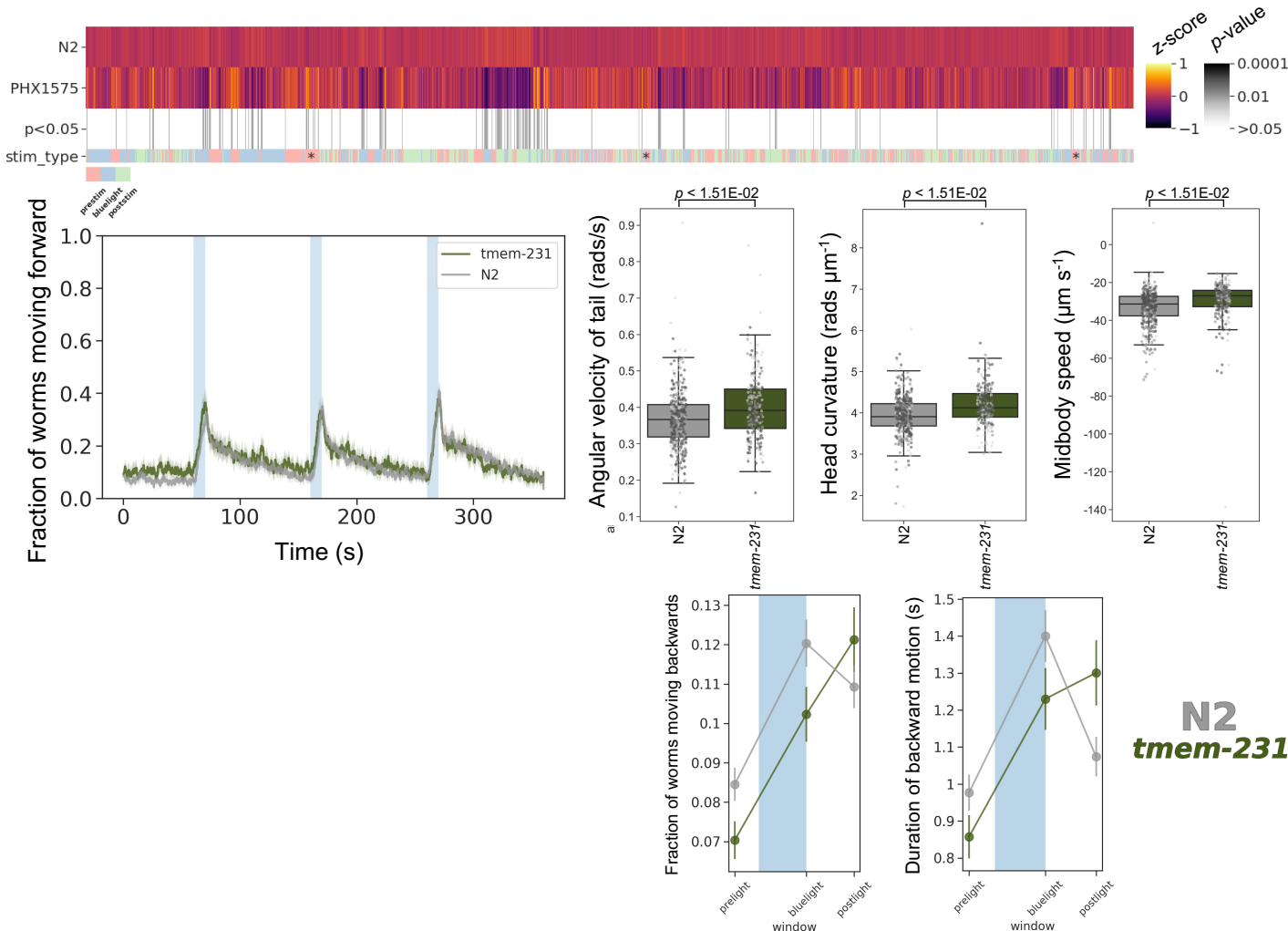
Associated disease(s):

Joubert syndrome 20, 614970 (AR); Meckel syndrome 11, 615397 (AR)

## Results

811/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): Slightly less active during peristimulus recording; increased head curvature and angular velocity of tail (during peristimulus only); increased speed upon stimulation with blue light; sustained backwards escape response to blue light



# *tub-1(syb1563)*

Strain name: PHX1563

Wild-type gene length: 1964bp

Mutation: 1319bp deletion at position 3 (exon 1 to beginning of exon 4)

## *C. elegans* Description

Function: Encodes TUBBY homolog and interacts with a 3-ketoacyl-coA thiolase, affecting lipid accumulation; involved in localisation of G-protein-coupled receptors in nonmotile cilia

Expression: Cytoplasm of ciliated cells

Previously reported phenotypes: Extended lifespan; increased fat content<sup>24-25</sup>

## Human Orthologs:

*TUB, TULP1*

Associated disease(s):

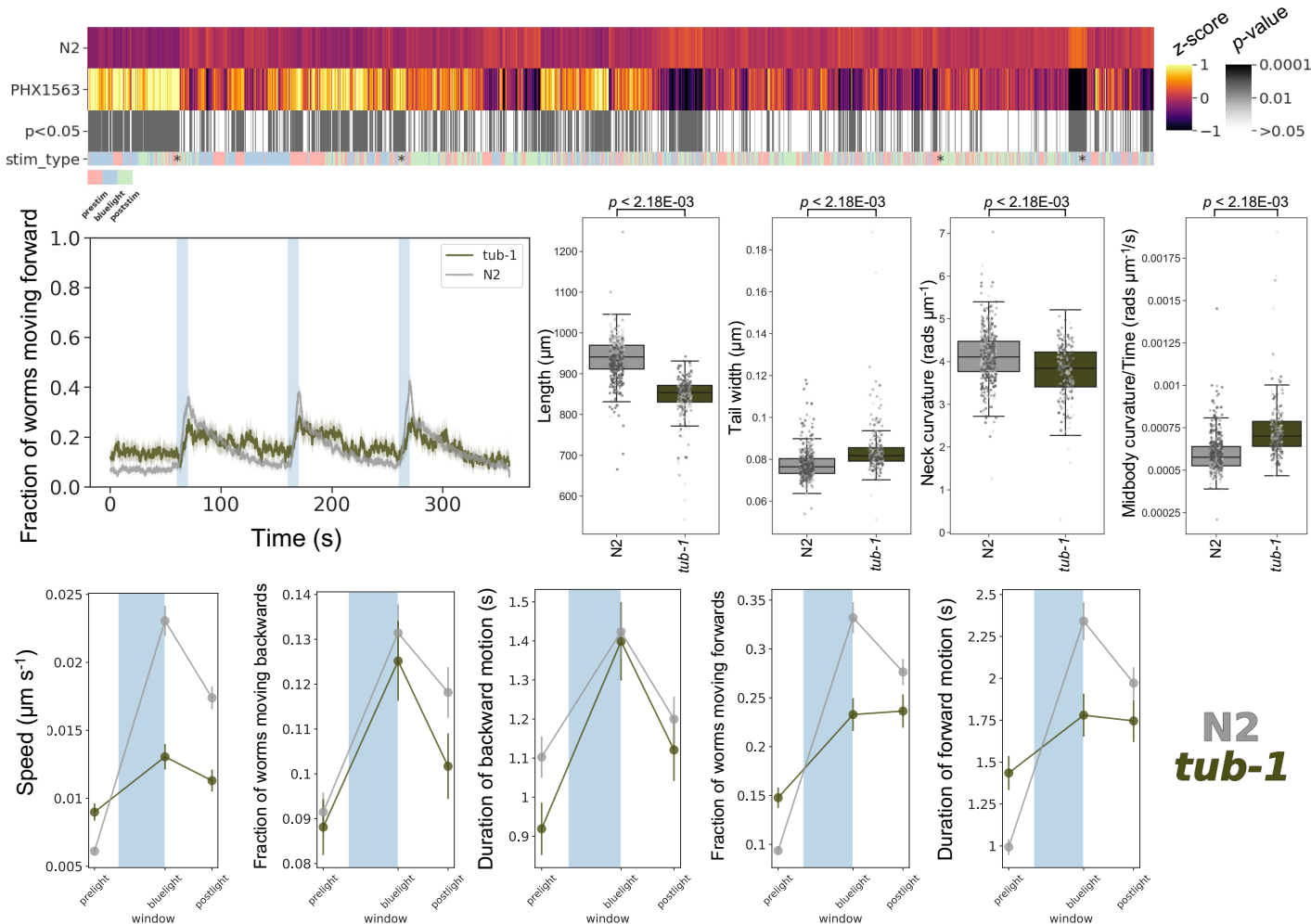
Retinal dystrophy and obesity, 616188 (AR); Leber congenital amaurosis 15, 613843 (AR);

Retinitis pigmentosa 14, 600132 (AR)

## Results

4567/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): Shorter and fatter; decreased curvature; deeper body bends (increased time derivative of curvature); hyperactive prior to simulation; decreased speed; attenuated, but sustained, sustained forward escape response; little difference in backwards motion/reversals



# *unc-25(syb1651)*

Strain name: PHX1651

Wild-type gene length: 1706bp

Mutation: 1304bp deletion at position 638 (exons 2-4)

## *C. elegans* Description

Function: Encodes glutamic acid decarboxylase (involved in GABA biosynthesis)

Expression: EF neuron and neurons

Previously reported phenotypes: GABA deficient; shrinker; defective defecation; irregular head thrashes; sensitive to PTZ, fluoxetine and aldicarb<sup>26-32</sup>

## Human Orthologs:

*GAD1*

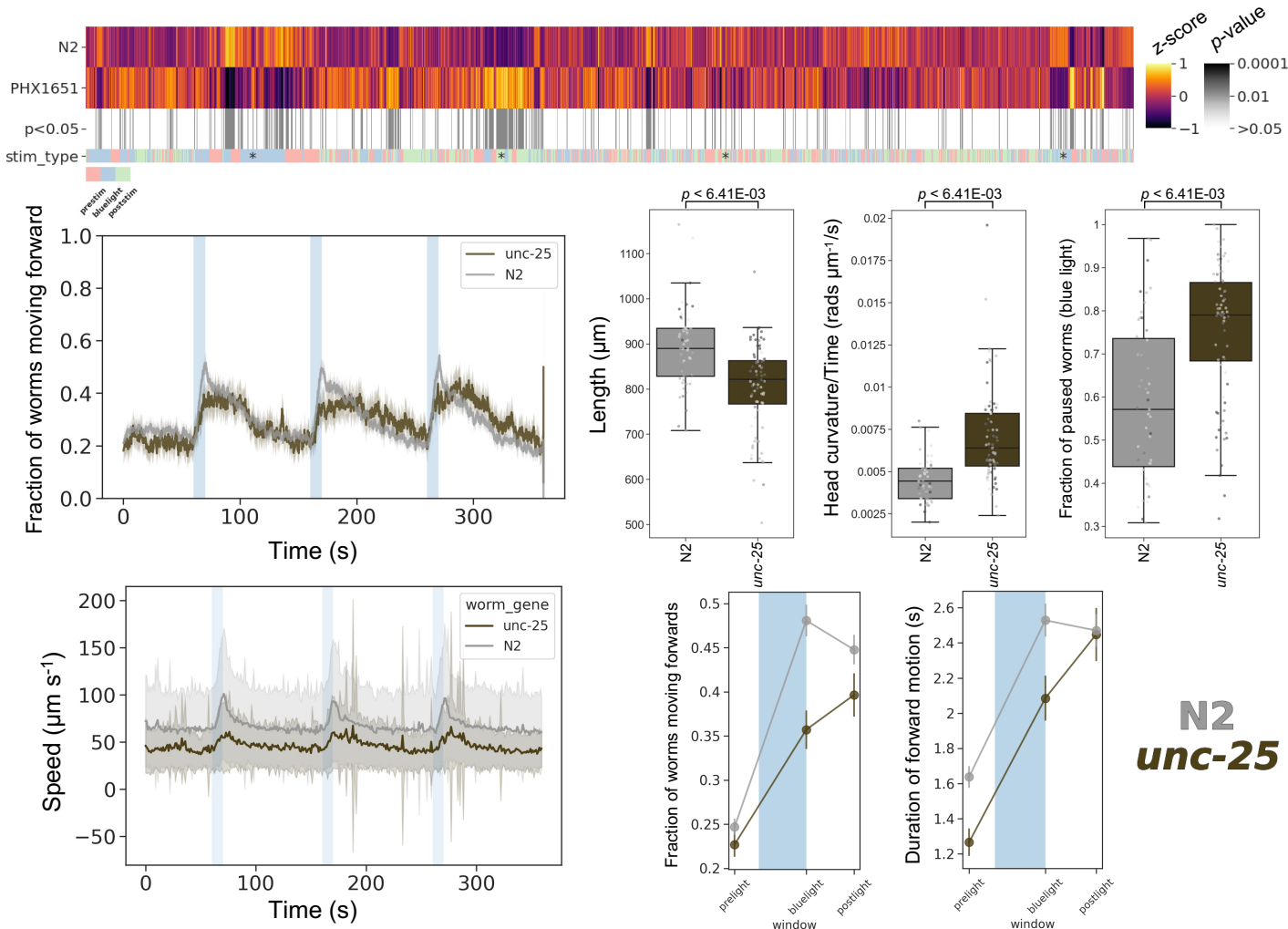
Associated disease(s):

Cerebral palsy, spastic quadriplegic, 1, 603513 (AR)

## Results

1956/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): Developmentally delayed (2 days); shorter; deeper sinusoidal bends (increased time derivative of curvature); less active and slower; delayed, but sustained forward escape response to blue light exposure



# *unc-43(syb2654)*

Strain name: PHX2654

Wild-type gene length: 1823bp

Mutation: 1246bp deletion at position 157 (exon 1 to beginning of exon 3)

## *C. elegans* Description

Function: Encodes CamKII-kinase; UNC-43 also interacts with the EGL-2 (ether-a-go-go potassium channel) to regulate it's activity

Expression: Neurons; oocytes; gonadal sheath cells; copulatory spicule

Previously reported phenotypes: Poor backing; slow moving; shrinker; defective egg laying; body wall muscle variant; sensitive to PTZ and aldicarb<sup>30,33-36</sup>

## Human Orthologs:

*CAMK2A*; *CAMK2B*

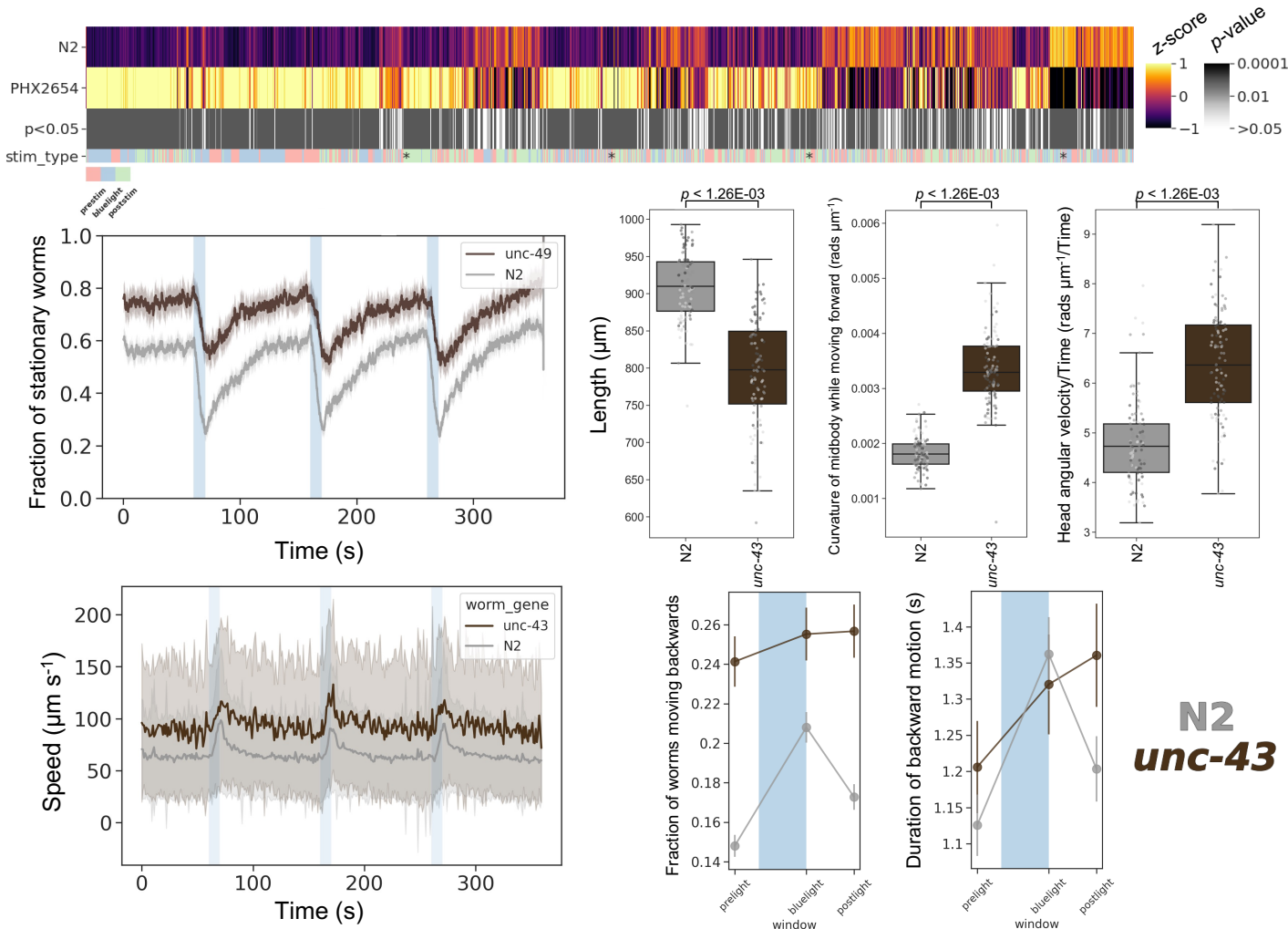
Associated disease(s):

Intellectual disability, 53, 617798 (AD); Intellectual disability, 54, 617799 (AD)

## Results

6858/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): Small; increased curvature and head foraging while moving (time derivative of angular velocity); hyperactive; increased speed; increased and sustained escape response to blue light stimulation



# *unc-49(syb2856)*

Strain name: PHX2856

Wild-type gene length: 8713bp

Mutation: 7765bp deletion at position 567 (9 exons)

## *C. elegans* Description

**Function:** Encodes subunits of heteromeric GABA receptor; UNC-49 is also required for postsynaptic GABA responsiveness

**Expression:** Expressed in anal sphincter muscle; body wall musculature; dorsal nerve cord; nerve ring and ventral nerve cord

**Previously reported phenotypes:** Decreased backing and thrashing; asymmetric body bends; small; shrinker; hypersensitive to fluoxetine and PTZ; resistant to muscimol<sup>12,17-18,28,30-32,37-38</sup>

## Human Orthologs:

*GABRB1; GABRB2; GABRB3*

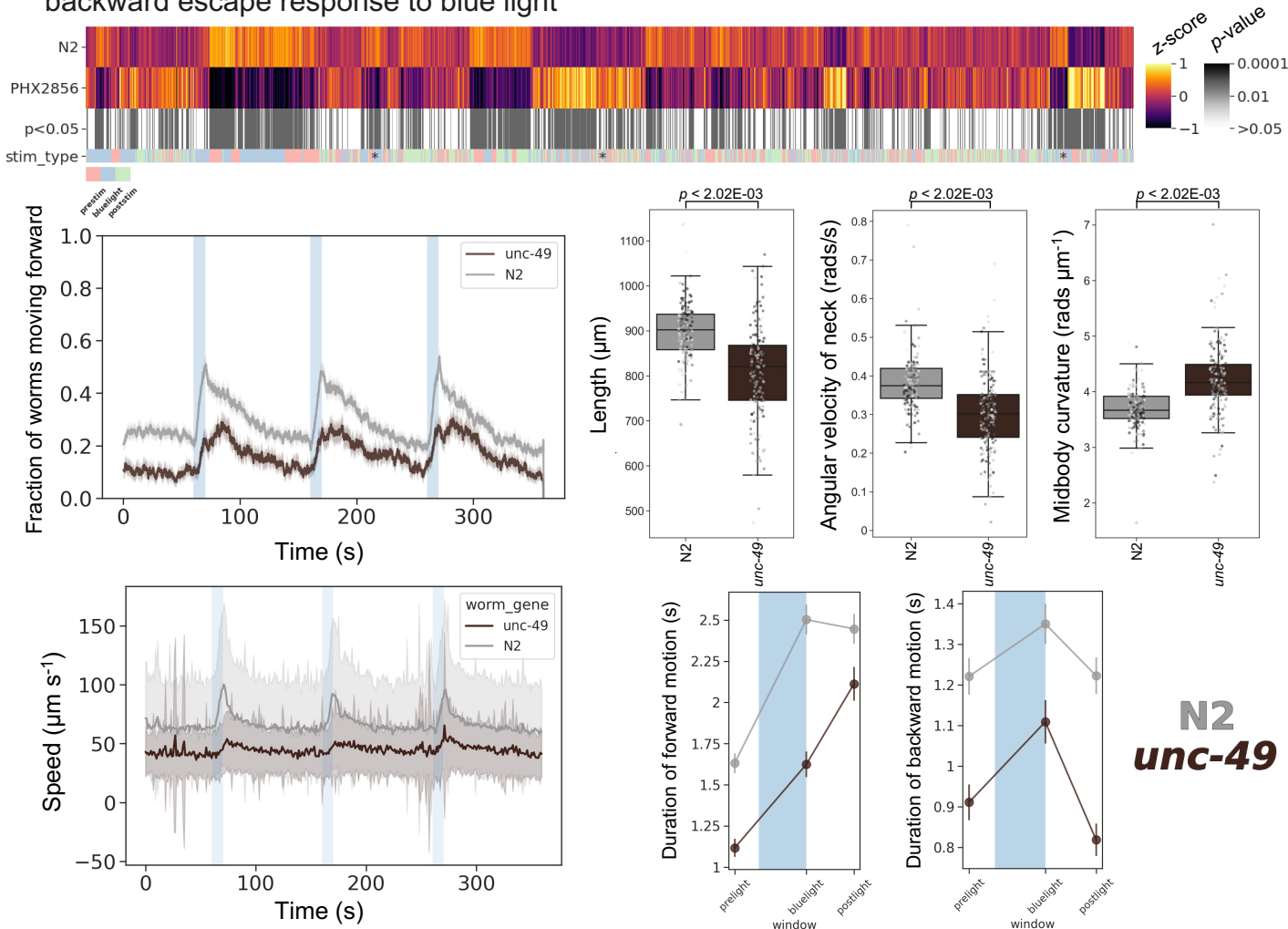
**Associated disease(s):**

Epileptic encephalopathy, early infantile, 43 and 45; Epileptic encephalopathy, infantile or early childhood, 2; Childhood absence epilepsy; Early onset epileptic encephalopathy

## Results

4799/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

**Key phenotype(s):** Less active; smaller; decreased angular velocity of neck; increased curvature of midbody; decreased speed; sustained forward escape response, but attenuated backward escape response to blue light



# *unc-77(syb1688)*

Strain name: PHX1688

Wild-type gene length: 2046bp

Mutation: 1407bp deletion at position 8 (first 4 exons of NM171353.4)

## *C. elegans* Description

Function: Cation-leak channel; interacts with *unc-80*, *unc-79* and *nca-2*

Expression: Cholinergic neurons; nerve ring; pharyngeal neurons and tail ganglion

Previously reported phenotypes: Decreased backing and increased sinusoidal movement<sup>15</sup>

## Human Orthologs:

*NALCN*

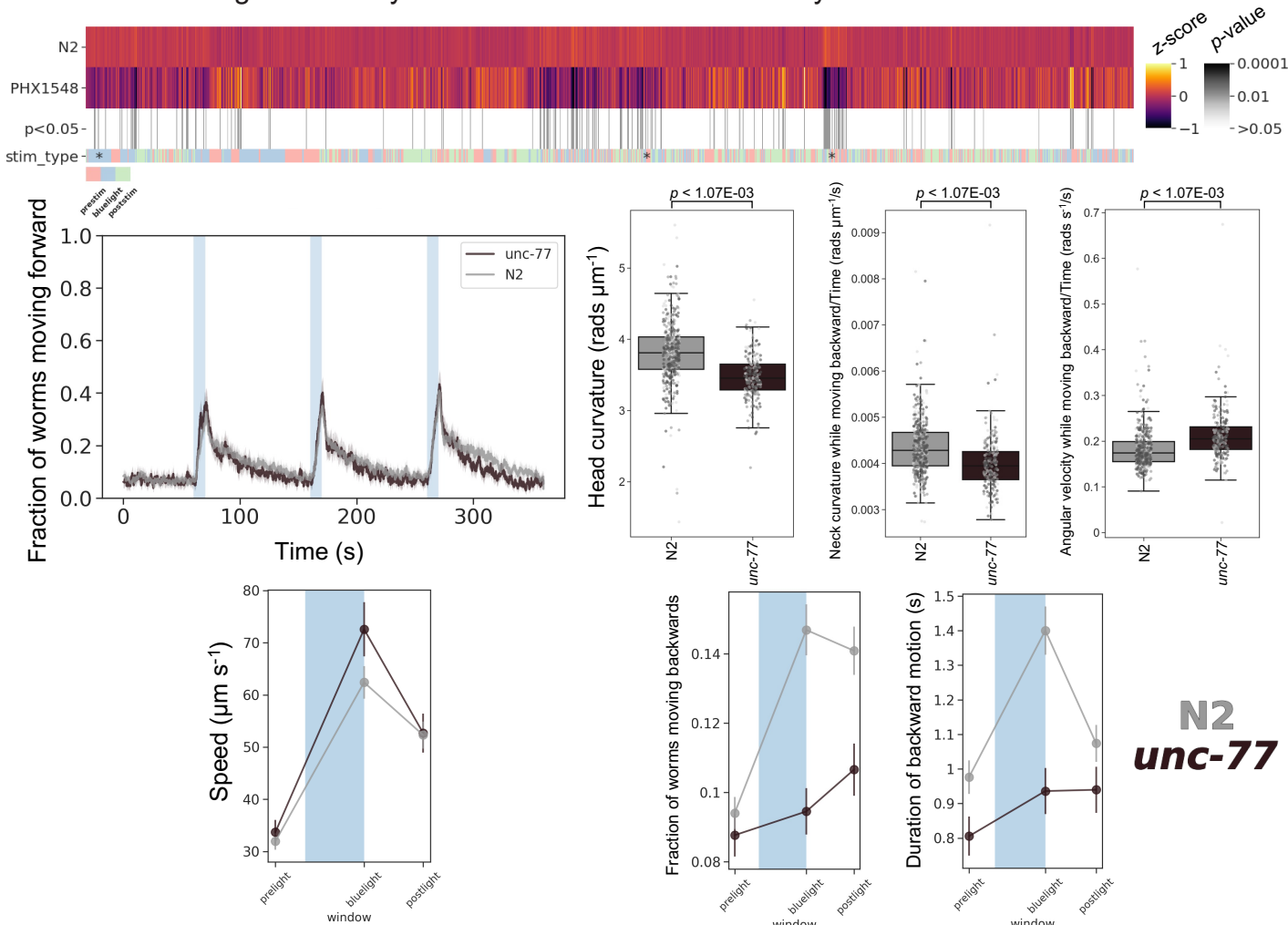
### Associated disease(s):

Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 (AD); Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 (AR)

## Results

1011/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): Decreased curvature of head/neck; stronger increase in speed in response to blue light; defective backing response, characterised by: decreased rate/duration of reversals, low frequency back bends (time derivative of curvature) and increase in time derivative of angular velocity related features across the body





# *unc-80(syb1531)*

Strain name: PHX1531

Wild-type gene length: 1806bp

Mutation: 1367bp deletion at position 60 (second half of exon 1 to middle of exon 4)

## *C. elegans* Description

Function: Cation-leak channel; interacts with *unc-77*, *unc-79* and *nca-2*

Expression: Cholinergic neurons; excretory canal; hermaphrodite distal tip cell; non-striated muscle and somatic nervous system

Previously reported phenotypes: Sinusoidal, coiling and locomotion phenotypes; fainting in liquid; sensitivity to halothane, isoflurane and ethanol<sup>15,39-40</sup>

## Human Orthologs:

*UNC80*

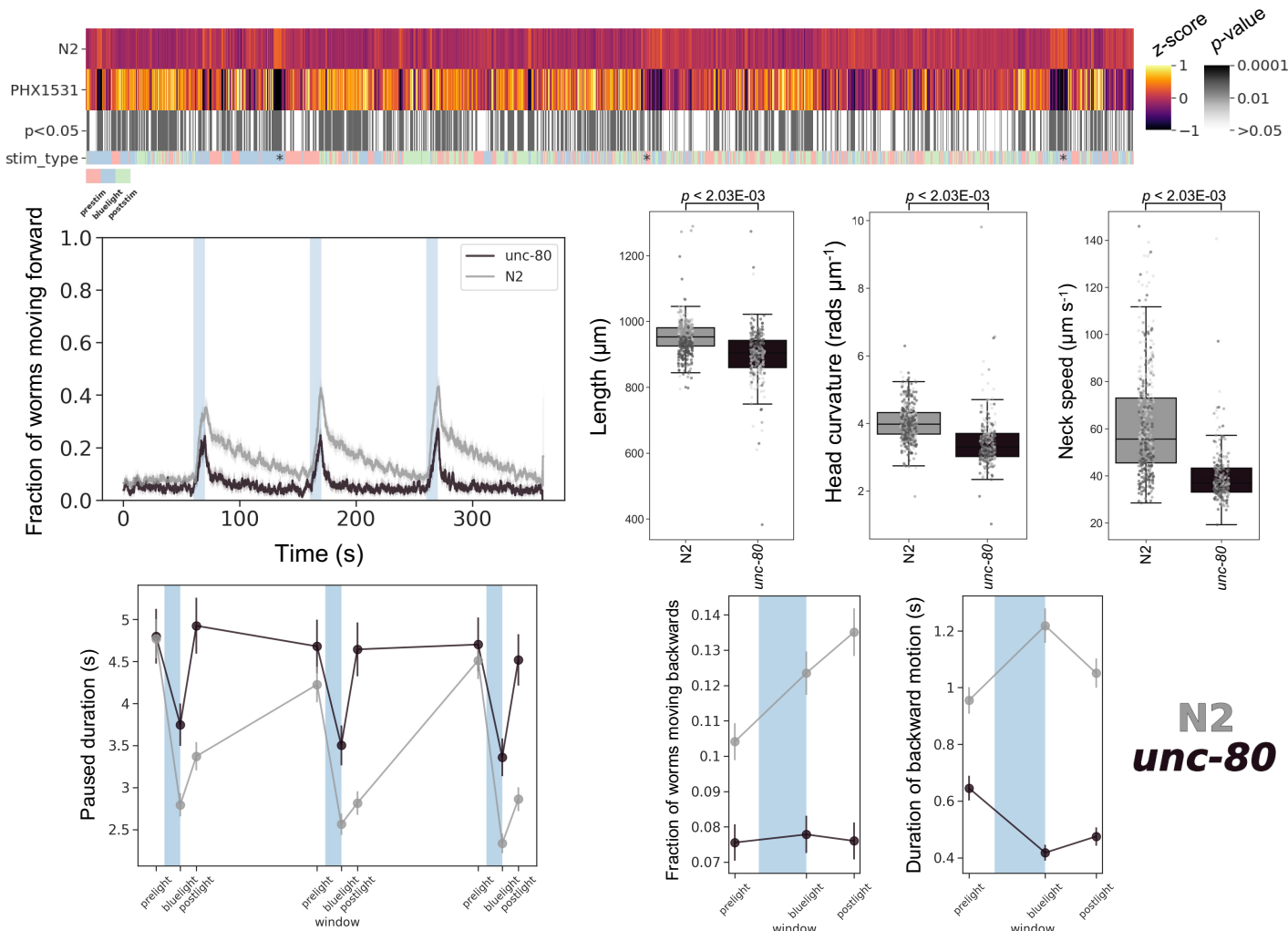
Associated disease(s):

Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801 (AR)

## Results

4868/8289 significant features vs N2 ( $p < 0.05$ , block permutation t-test, 10000 permutations)

Key phenotype(s): Shorter; decreased curvature and speed; less active; attenuated blue light response (e.g., lack of backward escape response); and fainting (increased pausing) following cessation of blue light stimulation





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