Relating Genomic and Phenotypic Differences Between Brown Bears and Polar Bears

Rosie DiDonato, James Kelley and Andrey Grigoriev
Grigoriev Lab

RUTGERS UNIVERSITY | CAMDEN

Intro

- Polar bears are among the species facing possible extinction in the near future, while brown bears are thriving
- Polar bears and brown bears are very closely related
- Random mutations and traits not advantageous in temperate conditions were advantageous to surviving in Arctic conditions and became fixed
- Polar bears are larger than brown bears (can be seen in figure 1)
- Other phenotypic differences include head shape, fur color, and body type
- Purpose of study:
- Identify differences (variants) between the genomes of the two bear species
- Annotate genes affected by these variants to find how they may affect certain features of the bears including
- Phenotypic differences
- Environmental adaptations

Figure 1 shows the comparison of a polar bear and brown bear.

Figure 2 shows the comparison of the head of the polar bear (left) and brown bear (right).

References

1. Koscielny, G., et al. (2014). "The International Mouse Phenotyping Consortium Web Portal, a unified point of access for knockout mice and related phenotyping data." **42**(D1): D802-D809.

2. Rappaport, N., et al. (2013). "MalaCards: an integrated compendium for diseases and their annotation." **2013**.

Background

- An allele refers to one or more alternative forms of one gene that occur due to a mutation
- A fixed allele is when that variant is the only variant that exists for that gene
- Homozygous genes are two genes with two identical alleles
- Heterozygous genes are two genes with different alleles
- In this study, we analyzed the effects of structural variants and indels
- Indels: small insertion or deletion less than 50 base pairs
- Insertion: type of mutation where genetic material is added to the genome
- Deletion: type of mutation where genetic material is lost or deleted from the genome
- Variant of size: 50 base pairs such as deletions and insertions

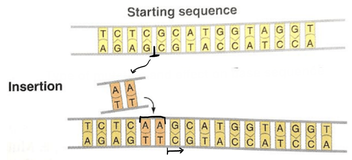

Figure 3 shows what an insertion looks like as ATAT is added into the sequence causing a mutation

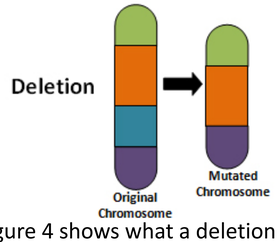

Figure 4 shows what a deletion looks like as genetic material is taken out from the original chromosome causing a mutation

Methods

- Genome Navigator was used in order to find fixed variants that overlapped coding regions of genes occurring in one species (brown bears or polar bears) that were not present in the other
- Resources used include:
- Genecards (which is mostly human data) used to find phenotypic differences among the two species https://www.genecards.org/
- MGI (Mouse Genome Informatics, mostly mouse data) used to find phenotypic differences among the two species http://www.informatics.jax.org/
- Google Scholar used to find primary references for papers describing gene function

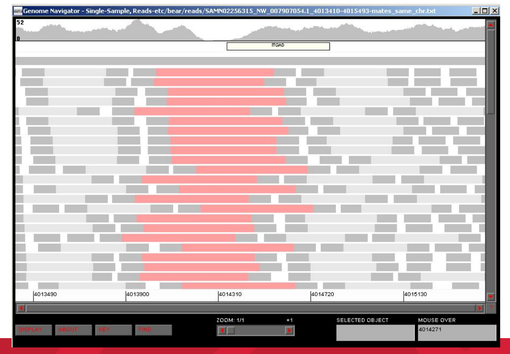

Figure 5 shows a deletion in Genome Navigator. The pink bars show evidence of a deletion in the DNA.

Results

- Body systems affected include:
- Head, body size, face morphology, skin, fur, internal diseases
- Genes related to fur and skin include: AP3D1, HAS3, ITGB2, TMEM79, and KMT2A (1)(2)
- Body size genes: ADD1, BMP7, COL17A1, CNTN1, MYBL1, RUNX3, WWP1, WWP2, and ZC3HC1 (1)(2)

Type of Variant	Gene	Affected	Affected Systems
DEL	BBS10	3' UTR	Body weight, leptin level (inhibits hunger)
INS	BMP7	CDS	Craniofacial and bone morphology, body size & weight
INDEL INS, INDEL	CAMLG	CDS, 5' UTR	Thermocyte and T-cell numbers
DEL	CDH16	3' UTR	B-cell number, IgG level
INDEL INS	CDK2AP	3' UTR	Snout size, forehead shape
INDEL INS	CHRM5	CDS	Circadian entrainment
INDEL DEL	COL17A	3' UTR	Hair pigmentation, body size
INDEL INS	HIVEP3	CDS	Skeletal physiology, bone mass
INDEL DEL	IDE	3' UTR	Cranium morphology, body weight
INDEL DEL	IFNAR1	3' UTR	Susceptibility to viral infection, B cell number, T cells
INS	KIF7	CDS	Head and face morphology
INS	LGR4	5' UTR	Body size, body weight, circadian rhythm
INS	MCM2	CDS	Hair pigmentation, body fat (affects subgroup)
INDEL INS	MGRN1	5' UTR	Hair pigmentation
INDEL DEL	MTHFD1	3' UTR	Head shape, head morphology
INDEL DEL	MYO5B	CDS	Body weight
INDEL INS	NR1H3	CDS	Circulating cholesterol level, susceptibility to bacterial
DEL	NR3C2	Exon	Body weight
INS	ORC6	RNA gene	Head size, skeletal
DEL	PARVB	3' UTR	Circulating cholesterol level, body weight
DEL	PDSS2	Exon	Circulating cholesterol level, weight, posture
INDEL INS, INDEL	PFKL	CDS	Glucose metabolism (reading frame restored)
DEL	PIGH	3' UTR	Circulating cholesterol level
5 INDELS	RAD18	3' UTR	Hair morphology, body weight, body fat amount
DEL	RECQL4	3' UTR	Hair morphology and pigmentation, body size, body fat
INDEL DEL	RGMA	CDS	Cranium morphology
INDEL DEL	RUNX3	3' UTR	Body size, height, weight, cranium morphology, skeletal,
INS, INDEL INS	SIRT2	3' UTR	Circulating cholesterol level
INDEL INS	SIRT3	3' UTR	Circulating cholesterol level
INDEL DEL	SLC1A1	5' UTR	Body fat amount
DEL	SLC29A3	5' UTR, RNA	Susceptibility to bacterial infection, hair pigmentation
INDEL DEL	TAF7	3' UTR	T cell number
INS	WDFY3	Exon	Head size
DEL	WDR35	3' UTR	Craniofacial & skeletal morphology
DEL	WDR47	5' UTR	Circulating cholesterol level, body weight

Table 1 shows genes related to systems in mouse and/or human studies, showing a small group of affected genes related to body size. Affected genes were found affected by structural variants and indels.

Acknowledgements

I would like to thank Dr. Grigoriev for his guidance throughout this research and James Kelley for his help and support with the data and research throughout this project.

RUTGERS