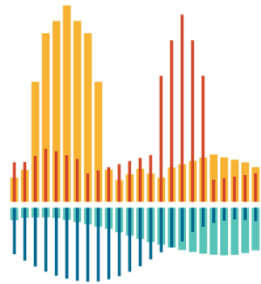


CRUNCHING NUMBERS: BIOINFORMATICS AND COMPUTATIONAL APPROACHES FOR GENOMIC STUDIES



INSTITUTE FOR
**COMPUTATIONAL
BIOLOGY**

October 14, 2016

William S. Bush
Assistant Professor
Epidemiology and Biostatistics
Institute for Computational Biology



SCHOOL OF MEDICINE

CASE WESTERN RESERVE
UNIVERSITY

My GENOMIC ANCESTRY

REGION	APPROXIMATE AMOUNT
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Europe 99%

■ Great Britain 64%

■ Europe West	17%
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Trace Regions 18%

■ Ireland	5%
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■ Scandinavia 4%

■ Finland/Northwest Russia	3%
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■ Italy/Greece 2%

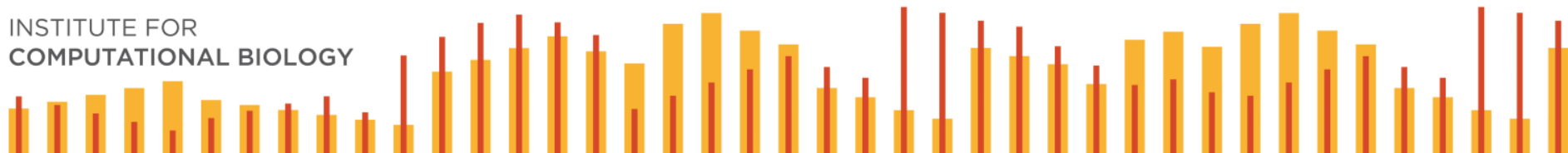
■ Europe East	2%
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■ Iberian Peninsula 2%

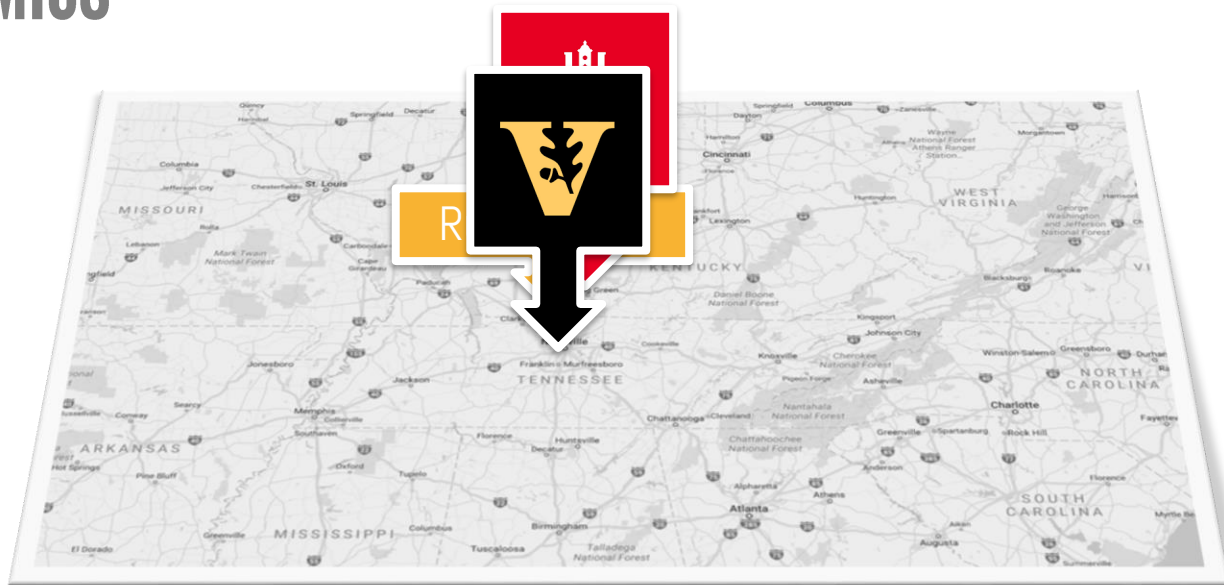
West Asia < 1%



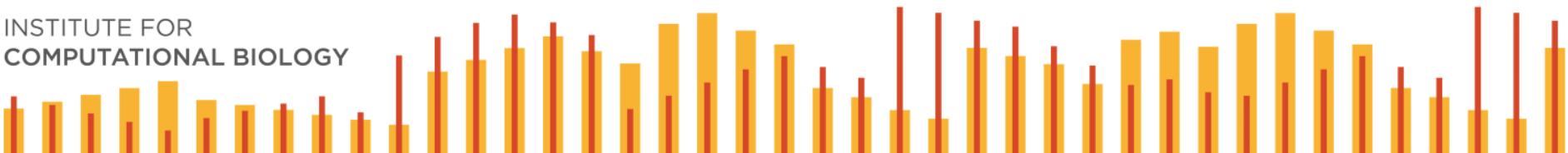
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My Journey to Genomics



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TRAIT/PHENOTYPE

BEHAVIOR

ENVIRONMENT

TISSUE/ORGAN FUNCTION

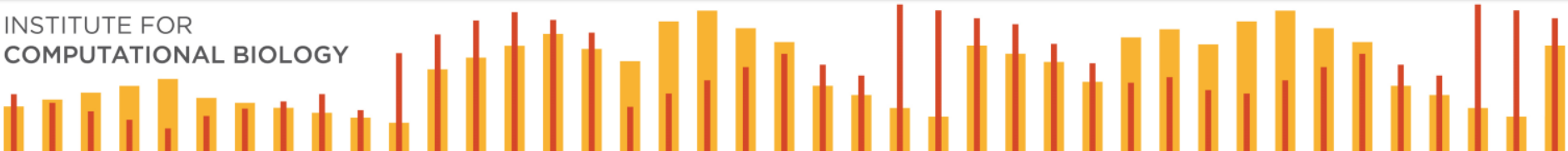
CELLULAR FUNCTION

PROTEOME

TRANSCRIPTOME

GENOME

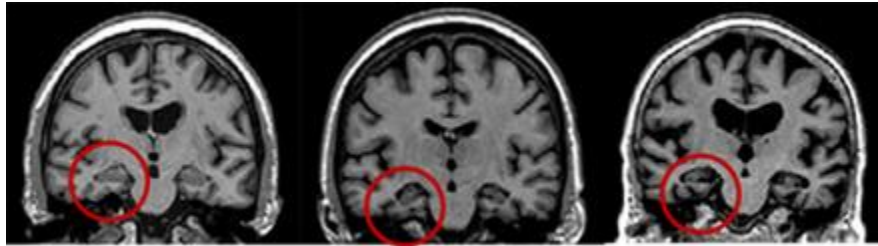
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TWO MANIFESTATIONS OF NEUROCOGNITIVE DECLINE

ALZHEIMER'S DISEASE (AD)

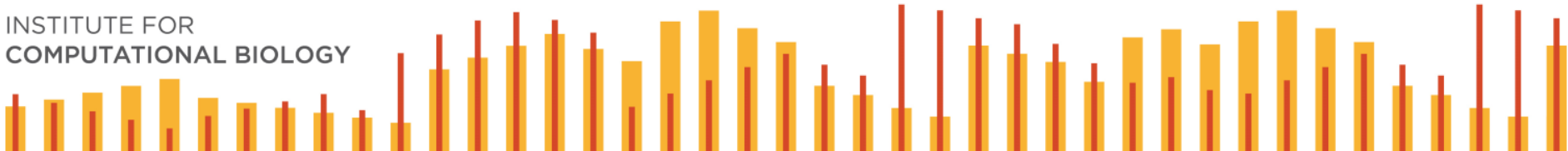
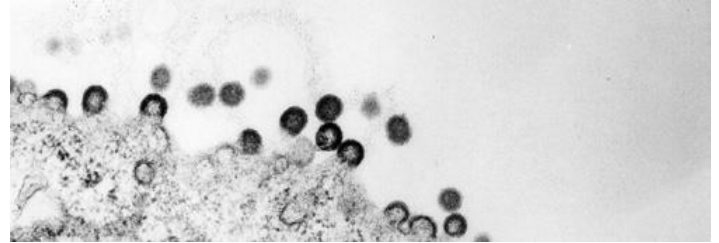
HIV ASSOCIATED NEUROCOGNITIVE DISORDERS (HAND)

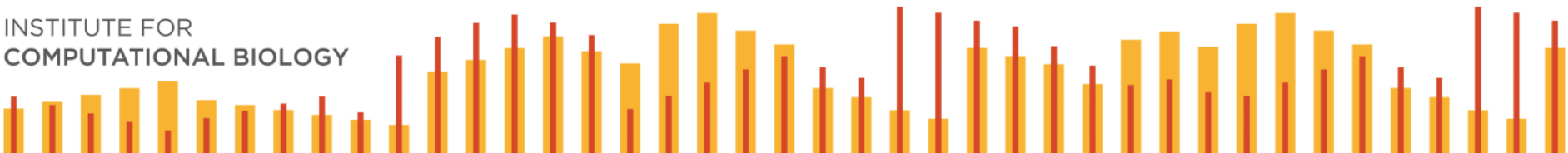
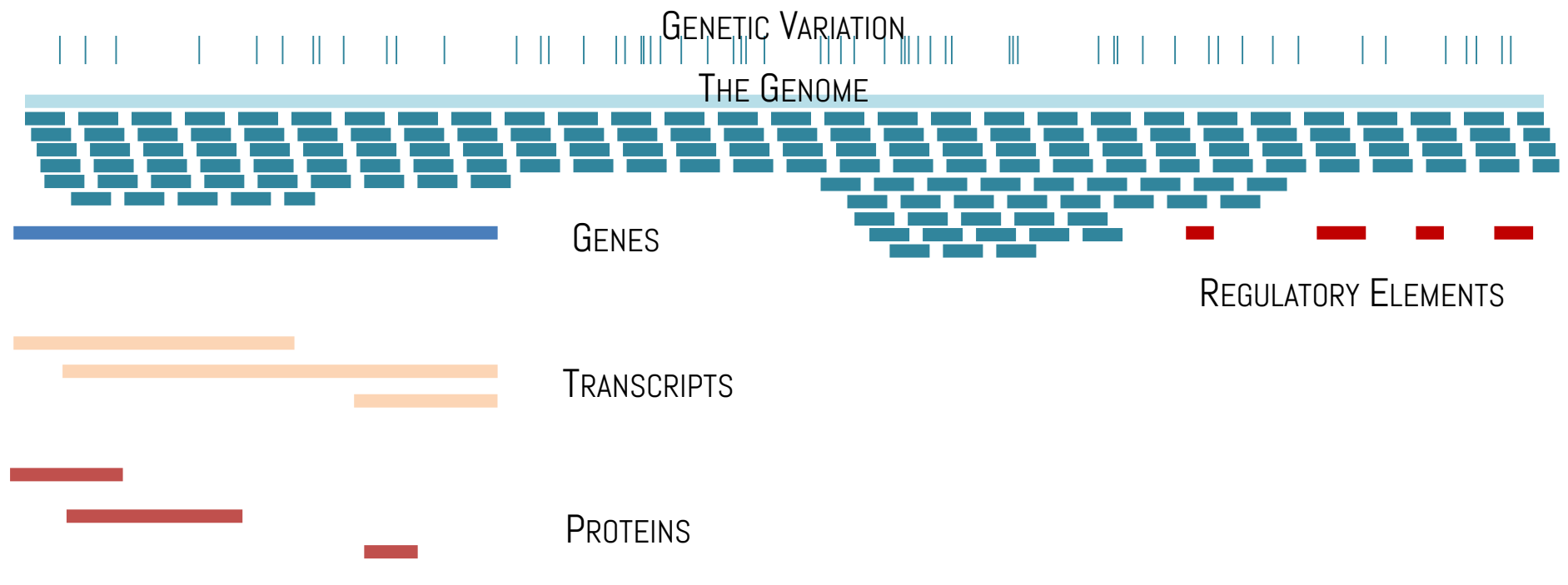


75 year old Control

75 year old MCI

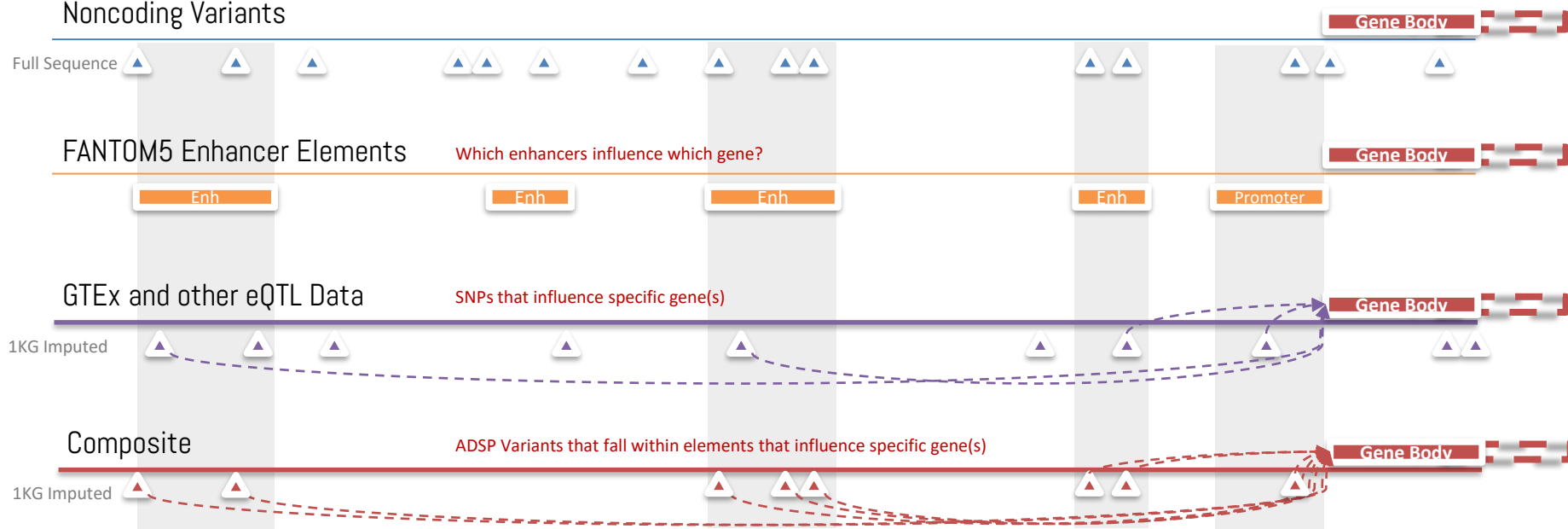
75 year old AD





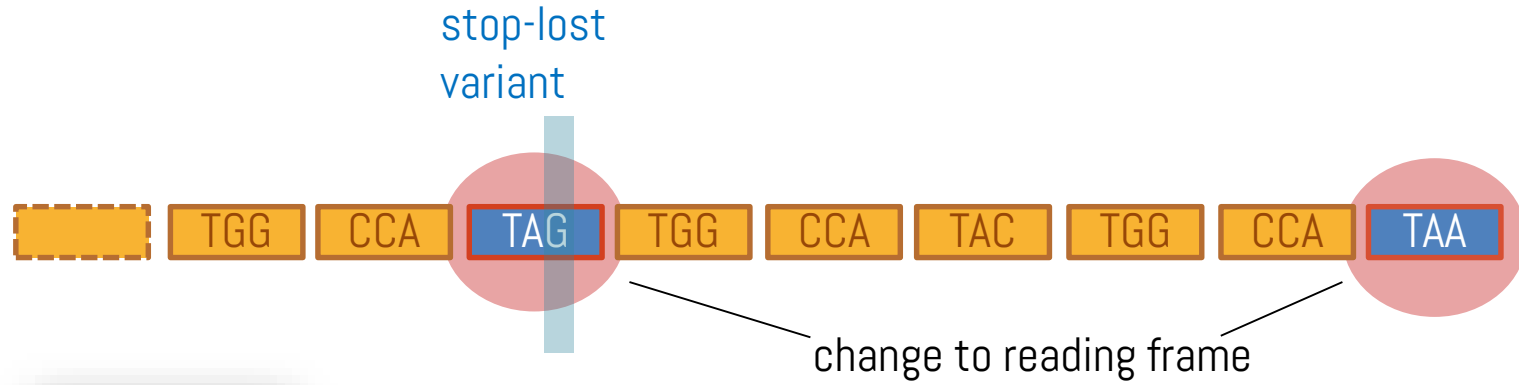
MAPPING REGIONAL VARIANTS TO GENES

Noncoding Variants

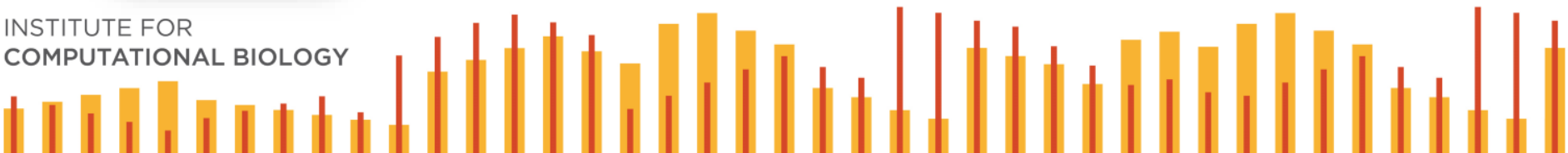


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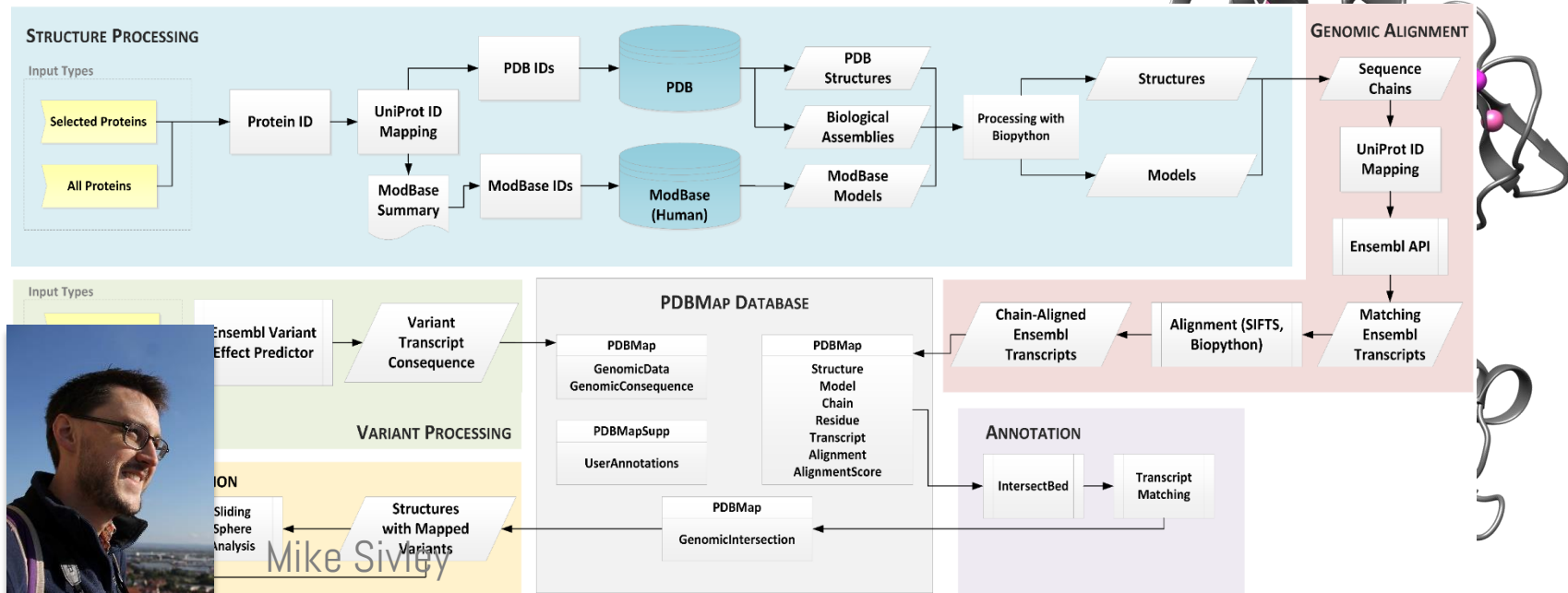
MAPPING VARIANTS THAT CREATE THE UNKNOWN



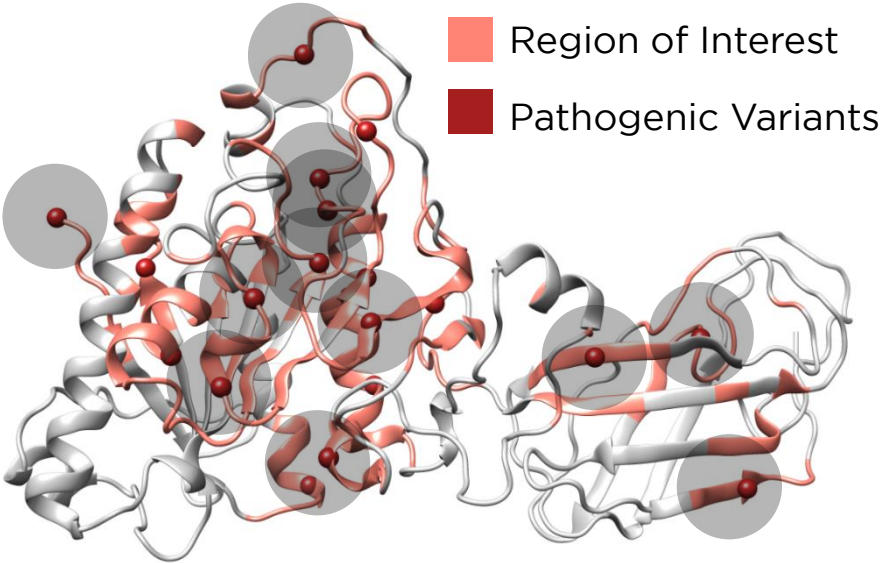
Mariusz
Butskiewicz



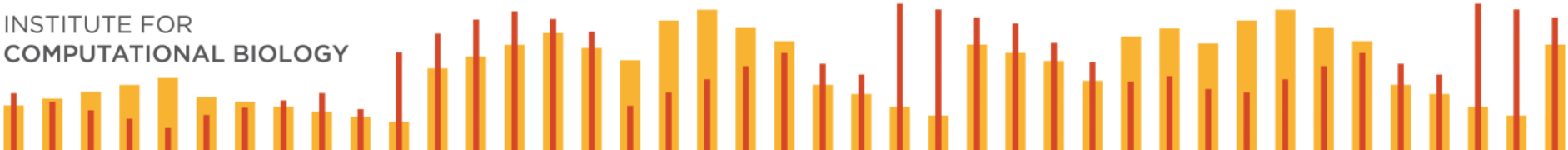
MAPPING VARIANTS INTO PROTEINS



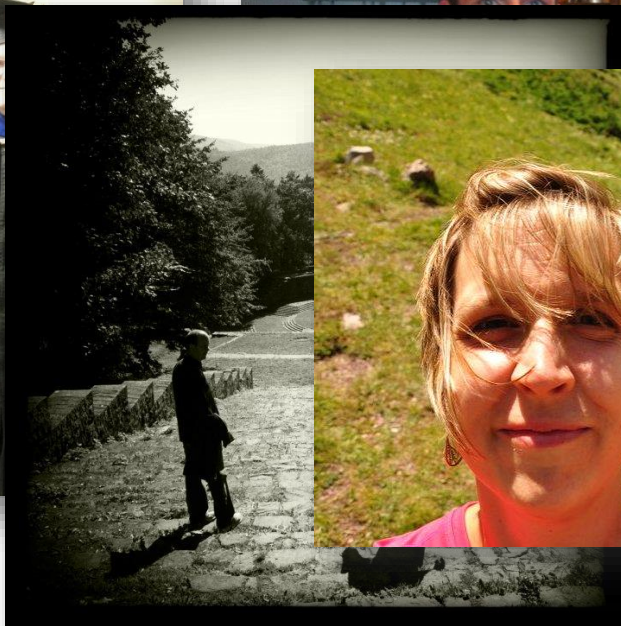
KNOWLEDGE-GUIDED ANALYSIS



Outcome	Total Count	P-values		
		Region of Interest	Outside Region	Whole Gene
HBA1C	20,093	0.8935	0.2492	0.2269
FBG	27,559	0.5302	0.1413	0.2492
CHOL	29,456	0.8233	0.4721	0.2676
FreeT3	6,412	0.2591	0.9142	0.6038
FreeT4	12,857	0.3126	0.5496	0.2201
HCO3	2,408	0.8202	0.2366	0.1201
HDL	29,597	0.0056	0.9589	0.0220
LDL	29,342	0.8854	0.7098	0.3216
TRIG	29,586	0.0766	0.1053	0.0010
TSH	2,650	0.6350	0.7079	0.5728



WORKING WITH COOL PEOPLE



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