

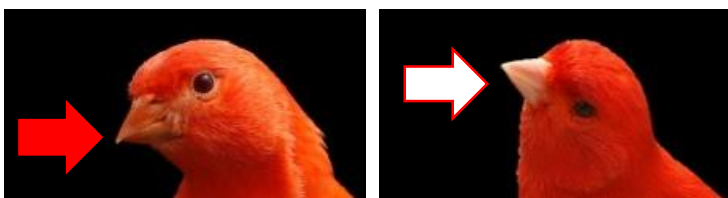


Through genetic mapping we singled out a **point mutation** affecting an **evolutionarily conserved** amino acid, which protein structural models and *in vitro* functional assays demonstrate it leads to **reduced rates** of carotenoid cleavage **activity of the *BCO2*** gene.

## Genetic basis of de novo appearance of canary bare-part coloration

Małgorzata Anna Gazda, CIBIO, Univeristy of Porto

### MODEL SYSTEM



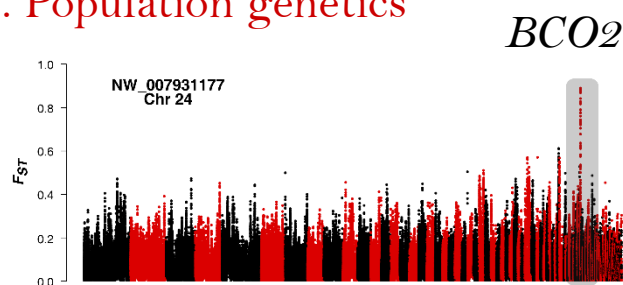
- Several decades ago, a canary with a red bill and legs appeared spontaneously in a colony
- Urucum canaries present a unique opportunity to identify locus that enables birds to express a conspicuous new ornamental color trait.

### CONCLUSIONS

- carotenoid-cleaving activity leads to coloration gain, modification, or loss
- interplay of the signaling benefits and physiological costs may be mediated by regulation of *BCO2*

### RESULTS

#### 1. Population genetics



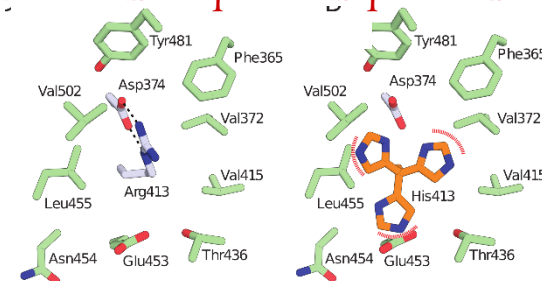
*BCO2* is a strong candidate gene based on population differentiation across the genome

#### 2. Evolutionary conservation

Canary wild type	P	R	A	F	P	R	R	F	V	L	P	L	D	V	D	S	D	T	P		
Canary urucum	.	.	.	.	.	.	H	.	.	.	.	.	.	.	.	.	.	.	.		
Chicken	A	.	T	.	.	.	.	.	.	.	.	.	.	.	.	.	K	.	N		
Rock pigeon	S	.	.	.	.	.	.	.	.	.	.	.	.	.	.	N	.	N	A		
Human	A	K	S	.	.	.	.	.	.	.	.	.	.	.	.	N	.	S	L	N	A
Cattle	A	.	N	S	.	.	.	.	.	.	.	.	.	.	.	L	G	N	L	N	A
Horse	A	K	S	.	.	.	.	.	.	.	.	.	.	.	.	H	.	S	L	.	A
Brown rat	A	K	S	.	.	.	.	.	.	.	.	.	.	.	.	I	.	S	V	G	A
Sperm whale	A	.	S	.	.	.	.	.	.	.	.	.	.	.	.	H	.	S	L	N	A
Chinese alligator	.	K	.	.	.	.	.	.	.	.	.	.	.	.	.	.	T	.	S	.	.

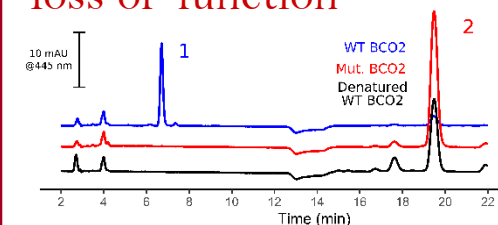
The mutation occurs in a highly conserved position across birds and vertebrates

#### 3. Mutation impact on protein activity



Change of aminoacid from Arg to His cause structural distortion of the protein leading to the loss of activity

#### 4. Functional test of mutated protein loss of function



- 1. Apocarotenoid
- 2. canthaxanthin substrate

Lysates containing the mutant *BCO2* did not produce apocarotenoids

CONTACT:

Email: m.gazda@cibio.up.pt Twitter: @GazdaMalgorzata

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