

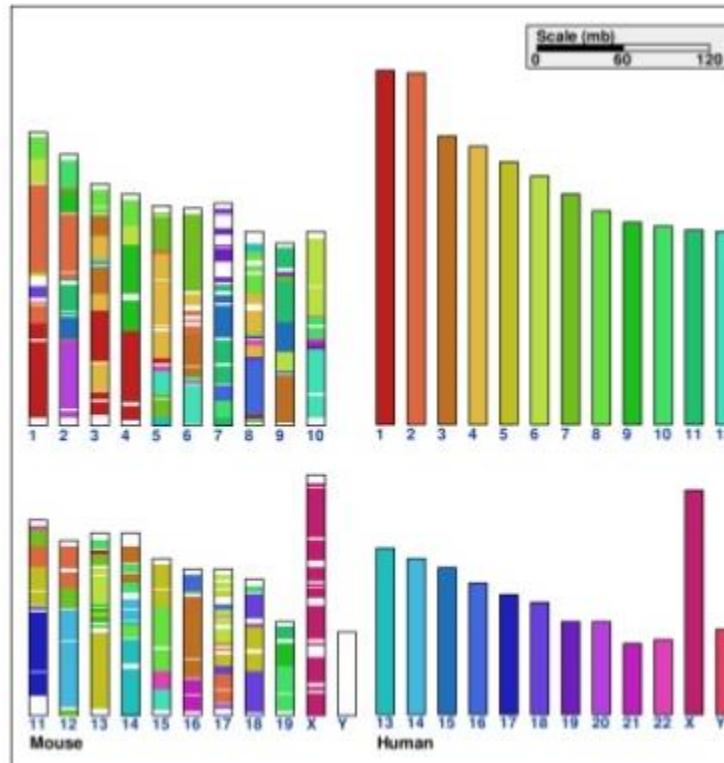
# Synteny Blocks

MPA-PRG: Programming in Bioinformatics

Exercise 8

# Synteny Blocks

- a group of consecutive genes that do not have any structural aberrations (insertions, deletions, translocations of other genes)



# Comparison of Synteny Blocks

- compare the chromosome set of two organisms
- find the synteny blocks
- compare the positions of synteny blocks
- infer the possible steps of chromosomal aberrations leading from an unknown ancestor to the current descendants
- comparison of synteny blocks within several organisms can give a rough picture of the genome of their mutual ancestor

# Sorting by reversals

- let  $\pi = \pi_1 \pi_2 \dots \pi_n$  be a permutation of  $n$  distinct numbers and  $1 \leq \pi_i \leq n$
- the reversal  $\rho = \rho(i, j)$  for  $1 \leq i < j \leq n$  applied to  $\pi$  reverses the values of  $\pi_i \pi_{i+1} \dots \pi_{j-1} \pi_j$  and thus transforms  $\pi$  into a permutation  
$$\pi \cdot \rho(i, j) = \pi_1 \dots \pi_j \pi_{j-1} \dots \pi_{i+1} \pi_i \dots \pi_n$$
- for example:  $\pi = 1 2 4 3 7 5 6$ , then  $\pi \cdot \rho(3, 6) = 1 2 5 7 3 4 6$
- the identity permutation  $I$  is the permutation where each  $\pi_i = i$  for  $1 \leq i \leq n$
- the distance  $d(\pi, I)$  between  $\pi$  and  $I$  is the minimum number of reversals  $\rho$  that transform  $\pi$  to  $I$

# Breakpoint Sort

- greedy algorithm
- minimize breakpoints between synteny blocks
- **adjacency** = a pair of adjacent elements that are consecutive
- **breakpoint** = a pair of adjacent elements that are not consecutive
  - 1 2 . 4 3
- block reversals are performed in such a way, that the number of breakpoints is reduced (or remains the same)
- each reversal removes at most 2 breakpoints
- number of reversals = hypothetical number of chromosomal changes

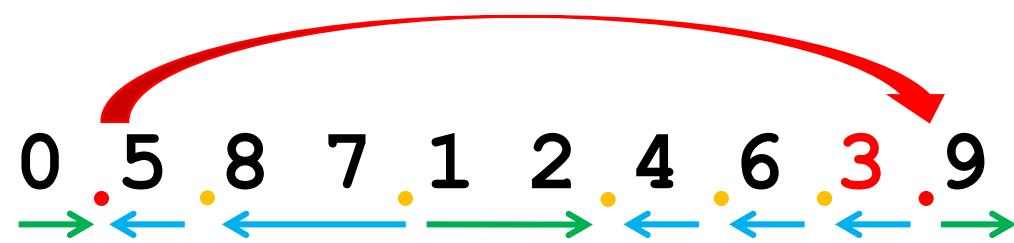
# Breakpoint Sort – Steps

- extend the permutation  $\pi_1 \dots \pi_n$  by  $\pi_0 = 0$  and  $\pi_{n+1} = n + 1$  on the ends,  $\pi_0$  and  $\pi_{n+1}$  never change their positions
- mark the ascending and descending parts of the permutation
- reverse the descending part that will lead to the largest reduction in the number of breakpoints

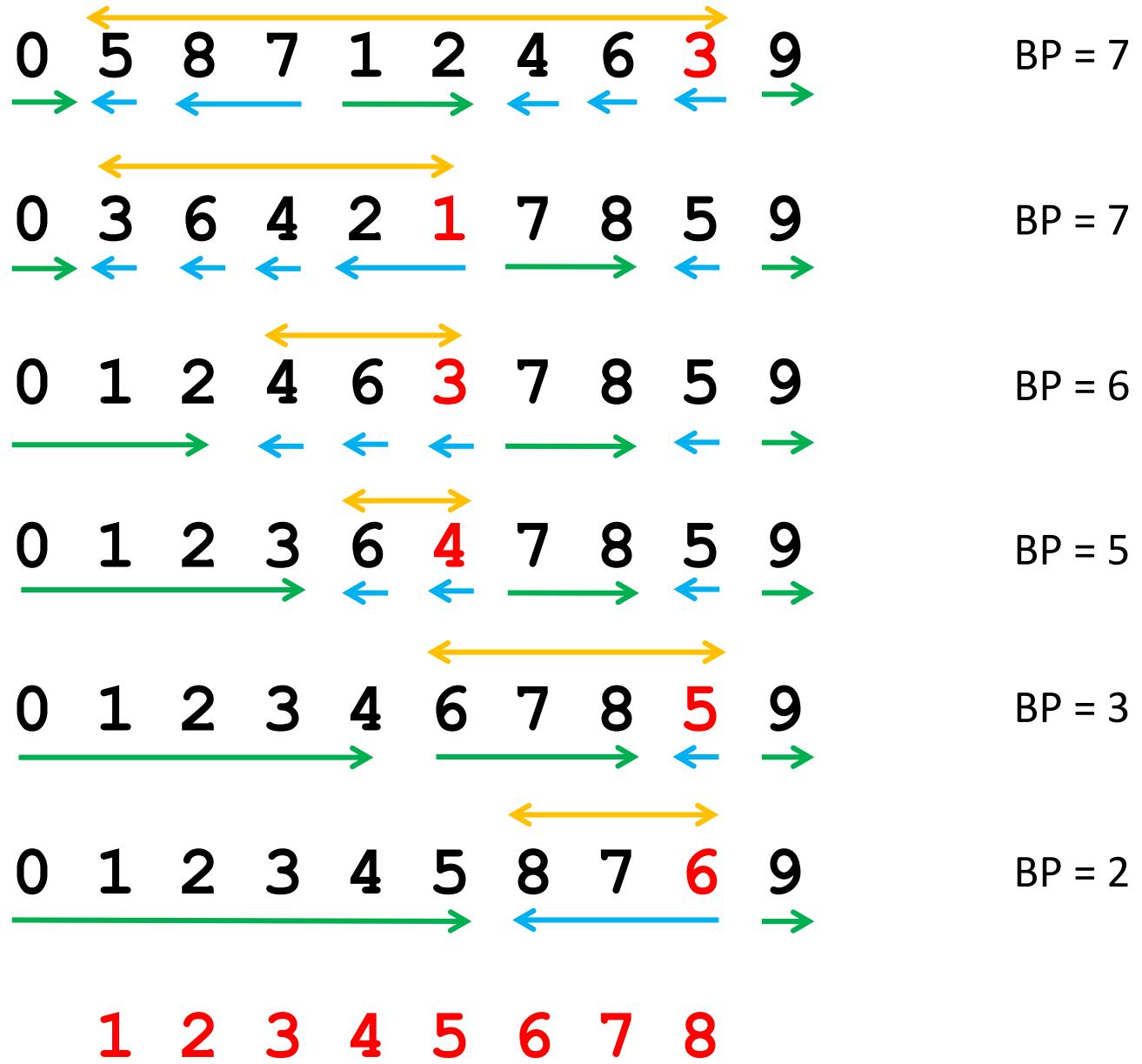


# Reversal

- find the descending part with the smallest value at the end
- reverse the region between the first breakpoint and the breakpoint following the selected descending part



# Example

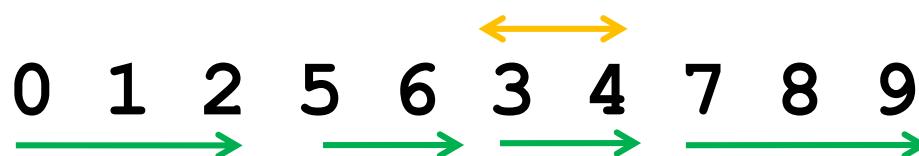


# No Descending Part

- reverse the entire block between the first and last breakpoint
- or the ascending part that starts with the value "end of sorted part" + 1

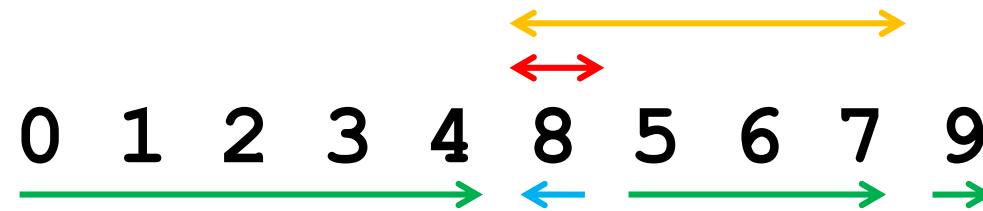


or



# One Descending Value in front of Sorted Part

- if the descending vector consists of one element and only the sorted part of the vector is in front of it, its reversal does not solve anything
- reverse the entire region between the first and last breakpoint



# Example

- sort permutation  $\pi = 5 \ 3 \ 2 \ 1 \ 8 \ 7 \ 4 \ 6$  using the Breakpoint Sort

# Tasks – The Breakpoint Sort

- **implement function** FindSorted ()
- **implement function** IndicateAscending ()
- **implement function** BreakpointSort ()

# Task 1

- in R, create a function `FindSorted()` to find an index, at which the unsorted part starts
- input:
  - a vector (permutation) of integers e.g. 0 1 2 3 6 7 4 5 8
- output:
  - an index, at which the unsorted part starts e.g. 5

**Hint:** Compare successively values of the permutation with an increasing number starting at zero (0, 1, 2, ...) and ending at length of the permutation - 1. The comparison ends when the value in permutation is not the same as the tested value or when the tested value is equal to the length of the permutation - 1.

# Task 2

- in R, create a function `IndicateAscending()` to mark ascending and descending parts of a permutation
- input:
  - a vector (permutation) of integers e.g. 0 4 5 3 2 1 6 7 8
- output:
  - a vector of zeros and ones, where ascending parts are marked by 1 and descending by 0 e.g. 1 1 1 0 0 0 1 1 1

**Hint:** Create an indication vector of the same length as the permutation containing only 0 values, and then set the first and last values to 1. The ascending parts of the permutation vector will be marked with 1 values in the indication vector. Create a loop that iterates through the permutation and if two values next to each other are ascending, i.e. the second is the first + 1, then the indication vector is set to 1 at the given indexes.

# Task 3

- in R, create a function `BreakpointSort()` to sort a permutation using Breakpoint Sort
- input:
  - a vector (permutation) of integers e.g. 5 1 4 3 7 8 9 2 6
- output:
  - sorted permutation of integers e.g. 1 2 3 4 5 6 7 8 9

**Hint:** Add marginal values to the permutation and the following steps are repeated in the loop:

- find the start of the unsorted region,
- mark ascending/descending parts,
- find the smallest value that is marked as descending part,
- reversal between the start of unsorted region and the smallest value marked as descending.

The loop ends when the permutation vector is sorted. Watch out for collision situations.