SplitSup User Guide

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Chapter 1

Introduction

This manual describes the use of SplitSup, C software that computes split scores as developed in [AKR16].

Under the general Markov model (GM) of base substitution, if DNA sequence data is generated on a tree T, then a matrix flattening of the pattern distribution for any split arising from an edge of T will be approximately of rank 4. Splits not arising from edges should have corresponding matrix flattenings of rank greater than 4. The software SplitSup returns split scores for checking if a matrix flattening is 'close' to rank 4 or not.

Possible split scores range from 0 (strongest support) to 1 (weakest support). The precise formula for the score, which depends on singular values of the matrix flattening, is given in [AKR16]. Theoretically, if a split occurs on a tree, the split score should approach zero as sequence length is increased.

For some purposes (e.g., mixture models, or a combined coalescent/substitution model as in [CK14]) researchers may wish to compute scores based on approximate rank other than 4. An option allows for this.

Current, only DNA sequence data is allowed. Modifications for amino acid sequences are possible, if a researcher has a compelling need for it.

Chapter 2

Using SplitSup

2.1 Input files

SplitSup requires two input files with the names infile and splitsfile:

File name	Contents
infile	DNA sequence data, in PHYLIP format
splitsfile	splits whose scores are to be computed

The data in infile must be sequential (not interleaved).

The file splitsfile is a text file, formatted as follows: The first line of the file is the number of splits for which scores will be computed. Each subsequent line specifies one of the splits, in the format

```
split-size elements-in-split-separated-by-spaces
```

Here the elements-in-split-separated-by-spaces is a list of elements of *one* of the partition sets in the split (the one of size split-size). For example, if splitsfile were

then SplitSup will compute split scores for two splits: a 5-split for which one of the partition sets contains taxa 1, 2, 3, 6, and 7 in infile and the other partition set contains the remaining taxa, and a 3-split for taxa 5, 6, and 7.

Note that splitsfile does *not* use the taxa names, but rather the order of the sequences in infile. For instance, if infile has, in order, sequences for human,

gorilla, chimp, orangutan, then human is taxon 1, gorilla is 2, chimp is 3, and orangutan is 4.

There is some ambiguity in naming splits. In analyzing a dataset with 10 taxa, the splits indicated by the lines

3 1 2 3 and 7 4 5 6 7 8 9 10

are identical.

2.2 Command line options

SplitSup may be used in two ways, though a variety of analyses fit into these schema. In the first way, splitsfile has a large number of splits for multiple scores computations. In the second way, splitsfile has only a single split, for a sliding window analysis.

The syntax for these analyses is

```
SplitSup [-rRANK] [-s -wWINDOW_SIZE -oOFFSET_SIZE -mMIN_NUMBER_SITES]
```

where square brackets indicate optional parameters. If a user calls the program with the basic command

SplitSup

then the two input files are read, and all split scores are written to the output file scoresfile, overwriting its previous contents. Details on the format of scoresfile are given below.

As an optional parameter, a user may choose the rank RANK underlying the score computation. For the GM model of DNA base substitution and its submodels such as GTR, the default value of RANK=4 is the appropriate one. For a 2-class mixture of the GM model, the rank of flattenings corresponding to true splits should be 8, so specifying -r8 is appropriate (see [AR06] for this and similar results). Underlying the software SVDquartets of [CK14, CK15] and its model of a combined coalescent and GTR+Γ+I substitution model are matrix flattenings for quartets which are of rank 10, so an -r10 option can be used to compute relevant scores with SplitSup.

The last set of optional parameters is used to select the sliding window analysis, in which split scores are computed for shorter segments of the alignments. For this, the splitsfile should contain only one split. Calling the program, for example, with

```
SplitSup -s -w1000 -o100 -m200
```

performs a sliding window analysis with a window size of 1000 bp and a slide step size (offset) of 100 bp, requiring a minimum of 200 non-gapped base pairs in any window for

the split score to be computed. The -s switch indicates that the sliding window analysis has been selected, and it must be followed by the three additional parameters.

To produce a sliding window analysis comparing several different splits requires running SplitSup several times with the \neg s option. For instance, in [AKR16] three splits were considered for some primate data: HC|GO, HG|CO, and HO|CG. A simple script file, with three versions of splitsfile was used, with each scoresfile copied before the next was produced.

2.3 Output file

SplitSup stores scores to an output file named scoresfile. The format of this file depends on whether the -s option for a sliding window analysis was invoked.

If -s was not invoked, so splitsfile potentially listed many splits, then scoresfile contains a list of scores for these splits. For instance if splitsfile had contents

```
12000
3 1 2 3
5 1 2 4 6 7
.:
(11998 more splits)
then scoresfile would be formatted as
12000
3 0.246620643974497
5 0.274404335917279
.:
(11998 more scores)
```

The first line of scoresfile indicates the number of scores that were computed. Subsequent lines in the file correspond to the ordered list of splits in splitsfile and have two entries: the size of the split and the value of the score.

If -s was invoked for a sliding window analysis, say with command

```
SplitSup -s -w10000 -o1000 -m500
and splitsfile
1
2 1 4
```

then scoresfile will be formatted as

```
:
169001
        638
               623
                     0.000076033553543
170001
        1631
               1568
                     0.000123768383328
171001
        2605
               2501
                     0.000765813771734
172001
        3600
               3451
                     0.000744102188514
173001
        4579
              4367
                     0.001116200230904
174001
               5284
        5542
                     0.001562678240580
175001
        6521
              6225
                     0.001635966713922
```

Each line of scoresfile has the following four entries: window-start-location, number-of-non-gapped-sites-in-window, number-of-constant-sites-in-window, and split-score-for-window. To interpret the first line shown above, for example, one sees that the sliding window started at site 169001 in the alignment, had 638 non-gapped sites, 623 constant sites, and the score for the 2-split was 0.000076033553543.

If a window fails to have the minimum required non-gapped sites set by the -m switch, there will be no output to scoresfile for it.

2.4 Warning

The distributions of split scores of k-splits for differing k are not the same. Comparing splits of different sizes by their split scores requires great caution. See [AKR16] for details.

Chapter 3

Getting and citing SplitSup

3.1 Sample files

A selection of sample analysis files are included in the directory sampleFiles on GitHub for help in learning to use SplitSup.

3.2 Required C libraries

The source code and supporting C files needed to compile SplitSup are all available at GitHub. Future versions and updates will be uploaded there. To compile SplitSup, download the following files from the GitHub repository and place them in the directory where you will compile the software.

```
flatBinary.c
main.c
svdlib.h
main.h
svdutil.c
las2.c
svdlib.c
svdutil.h
```

The GSL (GNU Scientific Library) library is also required. It is freely available from https://www.gnu.org/software/gsl//gsl.html. Depending on the location of the GSL installation on the local machine, C compiler flags -I and -L are likely needed. In the example below, the -I and -L flags point the C compiler to subdirectories of /opt/local/ where the GSL files reside. These paths should be changed appropriately.

Simple bare-bones commands for compiling SplitSup are

```
gcc -I/opt/local/include -c main.c
gcc -I/opt/local/include -c flatBinary.c
gcc main.o flatBinary.o -L/opt/local/lib/ -lgsl -lgslcblas -o SplitSup
```

3.3 Pronunciation and citation

The name SplitSup stands for *split support*, and should be pronounced "Split Soup". (While the name SplitPsup was attractive, we did not want to encourage an erroneous interpretation of split scores as *p*-values.)

If you use SplitSup for your research, please cite [AKR16]. This citation will be updated once the paper is accepted.

Bibliography

- [AKR16] E. S. Allman, L. S. Kubatko, and J. A. Rhodes. Split scores: a tool to quantify phylogenetic signal in genome scale data. http://arxiv.org/abs/1608.00942, pages 1-14, 2016.
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- [CK15] J. Chifman and L. Kubatko. Identifiability of the unrooted species tree topology under the coalescent model with time-reversible substitution processes, site-specific rate variation, and invariable sites. J. Theor. Biol., 374:35 – 47, 2015.