

# ANEXOS

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## GLOSARIO

Avconv: Librería que permite la edición de video y audio.

Datasets: Estructura del HDF5 que contiene información, usualmente matrices de números.

Groups: Estructura del HDF5 que puede contener otros *groups* o *datasets*.

HDF5: 1. Hierarchical Data Format. Formato de archivo caracterizado por estructurar la información usando *groups* y *datasets*. 2. Librería que permite leer y crear archivos con formato HDF5.

Indels: Inserciones y deleciones.

Librería: Conjunto de scripts que componen un determinado programa.

Matplotlib: Librería de Python para la creación de distintos tipos de gráficas en 2D y 3D.

NumPy: Numeric Python. Librería para Python escrita en C, C++ y Fortran que contiene diversas funciones para la creación de matrices y cálculos matemáticos.

ACP: Principal Component Analysis. Es una técnica usada para reducir las dimensiones de un conjunto de datos, utilizada para facilitar la búsqueda de la causa de la variabilidad de los datos.

Python: Lenguaje de programación diseñado en 1991 por Guido Van Rossum.

Script: Archivo de texto plano que contiene las órdenes (código) para realizar alguna tarea. Pueden ser considerados mini programas.

SNP: Single Nucleotide Polymorphism. Cambio de un nucleótido en un punto del genoma. A veces los indels se engloba en este término.

Ubuntu: Sistema operativo libre basado en GNU/Linux.

Variables: En programación, un conjunto de caracteres (usualmente una palabra que recuerde su contenido) que contiene la información que se le haya asignado.

VCF: Variant Call Format. Formato de archivo que almacena información sobre SNP e indels de varias muestras de una determinada especie o variedad con respecto al genotipo de referencia.

## **FILTROS\_ACP.PY**

```
import h5py
import allel
import numpy as np
```

```
#For create the graphic
from draw_ACP import draw_ACP
import sklearn
from sklearn.cluster import KMeans
```

```
def open_HDF5(filename):
    h5_file = h5py.File(filename, 'r')
    return h5_file
```

```
def extract_genotype_data(h5_file):

    """Transform the genotype data in h5_file to a GenotypeArray"""

    genotypes = h5_file['calldata']['genotype']
    #genotypes = h5_file['calldata']['GT']
    return allel.model.GenotypeArray(genotypes)
```

```
def filter_snps_by_maf(genotype_array, max_freq=1):

    """Eliminates the SNP with a allelic frequencie bigger than max_freq"""

    af = genotype_array.count_alleles().to_frequencies()
    maf = np.amax(af, axis=1)
    is_polimorf = maf < max_freq
    return genotype_array[is_polimorf, :, :]
```

```
def calculate_total_alleles(genotype_array):

    """With the genotype array, guess the ploidy and the number of samples to
    calculate"""
    """how many alleles per SNP have the array"""

    g_ploidy = genotype_array.ploidy
```

```
g_samples = genotype_array.n_samples
return g_ploidy*g_samples
```

```
def filter_snps_by_missing_calls(genotype_array, num_al_snp, max_missing):
```

```
    """Eliminate SNP whith less missing data than max_missing"""
    """num_al_snp is the total number of alleles per SNP"""
```

```
    al = genotype_array.count_alleles()
    suma = np.sum(al, axis=1)
    min_all = num_al_snp-max_missing
    filt = suma > min_all
```

```
    return genotype_array[filt, :, :]
```

```
def filter_snps_by_min_calls(genotype_array, min_calls):
```

```
    """Eliminates SNP whith less data than min_calls"""
```

```
    al = genotype_array.count_alleles()
    suma = np.sum(al, axis=1)
    filt = suma >= min_calls
    return genotype_array[filt, :, :]
```

```
def filter_gn(gn, num_al_snp):
```

```
    """Eliminates the rows with only 2"""
    """This filter is requiered if you have any row whith all 2, because ACP falls"""
    """Works with ploidy==2"""
    suma = np.sum(gn, axis=1)
    div =suma/num_al_snp
    filt = div<1 #If all are 2, div will be 1.
    return gn[filt, :]
```

```
def filter_(genotype_array, filters=True, num_al_snp=None, max_missing=0,
min_calls=1):
```

```
    """Uses at least maf filter"""
    """If filters==2 filts with missing call, 3 with min calls, and 4 with both"""
```

```
    if filters:
        g_filt = filter_snps_by_maf(genotype_array)
```

```
    if filters==2:
```

```

g_filt = filter_snps_by_missing_calls(g_filt, num_al_snp, max_missing)
elif filters==3:
g_filt = filter_snps_by_min_calls(g_filt, min_calls)
elif filters==4:
g_filt = filter_snps_by_missing_calls(g_filt, num_al_snp, max_missing)
g_filt = filter_snps_by_min_calls(g_filt, min_calls)

return g_filt

def obtain_ACP_points(ACP):
    "Obtain axes X, Y, Z like the first, second and third components"

    X = []
    Y = []
    Z = []
    valores=ACP[0] #ACP[0] are point data, ACP[1] is the object
    for i in range(len(valores)):
    X.append(ACP[0][i][0])
    Y.append(ACP[0][i][1])
    Z.append(ACP[0][i][2])
    #i is the sample number
    #The third number is the component

    return X, Y, Z

def calculate_Kmeans(gn, n_clusters):

    KM_cluster = KMeans(n_clusters=n_clusters)
    KM_fit = KM_cluster.fit(gn)
    centr = KM_fit.cluster_centers_
    X, Y, Z = centr[0], centr[1], centr[2]

    return X, Y, Z

def main(filename=None, filters=None, max_freq=1):

    filename = '/home/felipe/Documentos/HDF5/ri1_call2d.HDF5'
    #filename = '/home/felipe/Documentos/HDF5/calldata_2d_apeki.HDF5'
    #filename = '/home/felipe/Documentos/HDF5/tom_call2d.HDF5'

    h5_file = open_HDF5(filename)
    genotype_array = extract_genotype_data(h5_file)
    num_al_snp = calculate_total_alleles(genotype_array)
    g_filt = filter_(genotype_array, filters=2, num_al_snp=num_al_snp,
max_missing=30)

```

```

#g_filt = g_filt[:450000, :, :]
gn = g_filt.to_n_alt()

gn = filter_gn(gn, num_al_snp)

#Deleting the variables with genotypes that we don't need, increases speed and
#the max number of snp that can calculate the ACP function

del(genotype_array)
del(g_filt)

ACP = allel.stats.decomposition.ACP(gn)

X, Y, Z = obtain_ACP_points(ACP)

#X, Y, Z = calculate_Kmeans(gn, n_clusters=5)

return X, Y, Z

if __name__ == '__main__':

    proj = main(filename = '/home/felipe/Documentos/HDF5/tom_call2d.HDF5')
    draw_ACP(proj)

```

**DRAW\_ACP.PY**

```

from matplotlib.figure import Figure
from matplotlib.backends.backend_agg import FigureCanvasAgg as FigureCanvas
from mpl_toolkits.mplot3d import Axes3D
import os

```

```

def draw_ACP(projections, color='blue', dir_='.', opt_ax = 'off'):
    X, Y, Z = projections

    fig = Figure()

    axes = fig.add_subplot(111, projection='3d', xticks=[], yticks=[], zticks=[])
    axes.scatter3D(X, Y, Z, color=color)
    axes.axis(opt_ax)

    for i, angle in enumerate(range(0, 360, 1)):
        fname = 'ril_comp_%04d.png' % (i)
        fpath = os.path.join(dir_, fname)
        fhand = open(fpath, 'w')
        if not opt_ax=='off':
            axes.set_xlabel('componente 1')
            axes.set_ylabel('componente 2')
            axes.set_zlabel('componente 3')
        axes.view_init(azim=angle)
        canvas = FigureCanvas(fig)
        canvas.print_figure(fhand)
        fhand.close()

```

## TIEMPOS FILTROS

|  |  |  |  |       |  |  |  |
|--|--|--|--|-------|--|--|--|
|  |  |  |  | Numpy |  |  |  |
|  |  |  |  |       |  |  |  |

|        | t(s)   | tiempo<br>antes<br>primer<br>filtro | filtro maf=1 | filtro<br>maaf=1+<br>miss=100 | filtro<br>maaf=1+<br>miss=100+<br>min calls=1 | filtro<br>maaf=1+<br>miss=100+<br>min<br>calls=1+gn |  |
|--------|--------|-------------------------------------|--------------|-------------------------------|---|---|--|
|        | t1     | 0,94                                | 0,745        | 0,745                         | 0,691   | 0,794   |  |
|        | t2     | 0,726                               | 0,701        | 0,693                         | 0,696   | 0,712   |  |
| ril    | t3     | 0,743                               | 0,684        | 0,703                         | 0,703   | 0,708   |  |
|        | t4     | 0,724                               | 0,687        | 0,687                         | 0,718   | 0,701   |  |
|        | t5     | 0,733                               | 0,679        | 0,7                           | 0,703   | 0,704   |  |
|        | tmedio | 0,7732                              | 0,6992       | 0,7056                        | 0,7022  | 0,7238  |  |
|        |        |                                     |              |                               |   |   |  |
|        | t1     | 0,759                               | 0,738        | 0,773                         | 0,737   | 0,751   |  |
|        | t2     | 0,73                                | 0,733        | 0,734                         | 0,732   | 0,748   |  |
| apeki  | t3     | 0,723                               | 0,701        | 0,738                         | 0,745   | 0,757   |  |
|        | t4     | 0,726                               | 0,769        | 0,743                         | 0,737   | 0,752   |  |
|        | t5     | 0,751                               | 0,711        | 0,732                         | 0,789   | 0,763   |  |
|        | tmedio | 0,7378                              | 0,7304       | 0,744                         | 0,748   | 0,7542  |  |
|        |        |                                     |              |                               |   |   |  |
|        | t1     | 4,45                                | 27,27        | 13,066                        | 12,595  | 11,926  |  |
|        | t2     | 6,224                               | 24,103       | 14,646                        | 12,134  | 12,23   |  |
| tomate | t3     | 4,49                                | 17,549       | 13,726                        | 12,966  | 11,972  |  |
|        | t4     | 4,491                               | 9,205        | 12,197                        | 12,181  | 12,894  |  |
|        | t5     | 4,416                               | 9,032        | 12,149                        | 11,882  | 12,167  |  |
|        | tmedio | 4,8142                              | 17,4318      | 13,1568                       | 12,3516                                       | 12,2378   |  |

|        |  | For y np.vstack   |         |
|--------|--|-------------------|---------|
| t(s)   | af   | af(sin np.vstack) | gn      |
| t1     | 1,298                                      | 0,91              | 0,943   |
| t2     | 0,925                                      | 0,894             | 0,912   |
| t3     | 0,891                                      | 0,903             | 0,905   |
| t4     | 0,903                                      | 0,91              | 0,91    |
| t5     | 0,888                                      | 0,915             | 0,901   |
| tmedio | 0,981                                      | 0,9064            | 0,9142  |
|        |  |                   |         |
| t1     | 170,599                                    | 1,337             | 2,837   |
| t2     | 166,062                                    | 1,339             | 2,955   |
| t3     | 167,672                                    | 1,451             | 2,822   |
| t4     | 167,939                                    | 1,329             | 2,959   |
| t5     | 168,082                                    | 1,369             | 2,853   |
| tmedio | 168,0708                                   | 1,365             | 2,8852  |
|        |  |                   |         |
| t1     | 16min+(parado a mano (ctrlC) a 16minutos)  | 30,973            | 31,51   |
| t2     | 90min+(parado a mano (ctrlC) a 90 minutos) | 28,51             | 31,949  |
| t3     |  | 25,253            | 30,676  |
| t4     |  | 19,113            | 33,041  |
| t5     |  | 19,404            | 34,977  |
| tmedio |  | 24,6506           | 32,4306 |

## TIEMPOS POR MUESTRAS



|            |    |      |     |     |      |      |      |  |
|------------|----|------|-----|-----|------|------|------|--|
| 100k SNP   |    |      |     |     |      |      |      |  |
| nºmuestras | 50 | 100  | 150 | 200 | 250  | 300  | 348  |  |
| t1(s)      | 9  | 17   | 26  | 32  | 43   | 51   | 62   |  |
| t2(s)      | 9  | 17   | 26  | 32  | 43   | 51   | 62   |  |
| t3(s)      | 9  | 18   | 26  | 32  | 44   | 51   | 63   |  |
| t4(s)      | 9  | 17   | 26  | 32  | 43   | 51   | 62   |  |
| t5(s)      | 9  | 19   | 26  | 32  | 44   | 52   | 62   |  |
| tmedio     | 9  | 17,6 | 26  | 32  | 43,4 | 51,2 | 62,2 |  |
| 150k SNP   |    |      |     |     |      |      |      |  |
| nºmuestras | 50 | 100  | 150 | 200 | 250  | 300  | 348  |  |
| t1(s)      | 11 | 24   | 36  | 45  | 61   | 71   | 87   |  |
| t2(s)      | 11 | 24   | 36  | 45  | 61   | 71   | 87   |  |
| t3(s)      | 11 | 24   | 36  | 45  | 60   | 71   | 86   |  |
| t4(s)      | 11 | 23   | 36  | 45  | 61   | 71   | 86   |  |
| t5(s)      | 11 | 24   | 36  | 45  | 61   | 71   | 86   |  |
| tmedio     | 11 | 23,8 | 36  | 45  | 60,8 | 71   | 86,4 |  |

TIEMPOS NPY HDF5

|                      |                           | ril.vcf     |                |
|----------------------|---------------------------|-------------|----------------|
| tamaño comprimido    | 504,5kB                   | nºmuestras  | 153            |
| tamaño descomprimido | 2,5MB                     | nºSNP       | 943            |
|                      |                           | vcf2npy (s) | vcfnp2hdf5 (s) |
|                      | t1                        | 1           | 1,335          |
|                      | t2                        | 0,144       | 0,774          |
| variants             | t3                        | 0,148       | 0,704          |
|                      | t4                        | 0,154       | 0,701          |
|                      | t5                        | 0,163       | 0,773          |
|                      | tamaño                    | 223 K       | 477K           |
|                      | tamaño comprimido(tar.gz) | 74K         | 151K           |
|                      | tamaño comprimido (gz)    | 75K         | 152K           |
|                      | t1                        | 3,581       | 0,707          |
|                      | t2                        | 0,153       | 0,721          |
| calldata             | t3                        | 0,149       | 0,729          |
|                      | t4                        | 0,148       | 0,744          |
|                      | t5                        | 0,143       | 0,73           |
|                      | tamaño                    | 6,4M        | 1,5M           |
|                      | tamaño comprimido(tar.gz) | 438K        | 1,5M           |
|                      | tamaño comprimido (gz)    | 437K        | 1,5M           |
|                      | t1                        | 3,445       | 0,707          |
|                      | t2                        | 0,183       | 0,701          |
| calldata_2d          | t3                        | 0,153       | 0,719          |
|                      | t4                        | 0,15        | 0,759          |
|                      | t5                        | 0,152       | 0,756          |
|                      | tamaño                    | 6,4M        | 1,5M           |
|                      | tamaño comprimido(tar.gz) | 437K        | 1,3M           |
|                      | tamaño comprimido (gz)    | 437K        | 1,3M           |
|                      |                           |             |                |

|  |   |      |      |
|--|---|------|------|
|  | suma del tamaño de los 3 ficheros                     | 13M  | 3,5M |
|  | tamaño de los 3 ficheros comprimidos(tar.gz)          | 948K | 2,9M |
|  | suma del tamaño de los tres ficheros comprimidos (gz) | 948K | 2,9M |

|                      |                           |              |                |
|----------------------|---------------------------|--------------|----------------|
|                      |                           | tomate_apeki |                |
| tamaño comprimido    | 9,7MB                     | nºmuestras   | 12             |
| tamaño descomprimido | 35,6MB                    | nºSNP        | 41682          |
|                      |                           | vcf2npy (s)  | vcfnp2hdf5 (s) |
|                      | t1                        | 6,528        | 1,211          |
|                      | t2                        | 0,15         | 1,221          |
| variants             | t3                        | 0,151        | 1,232          |
|                      | t4                        | 0,149        | 1,243          |
|                      | t5                        | 0,146        | 1,271          |
|                      | tamaño                    | 9,6M         | 4,9M           |
|                      | tamaño comprimido(tar.gz) | 2,7M         | 4,5M           |
|                      | tamaño comprimido (gz)    | 2,7M         | 4,5M           |
|                      | t1                        | 14,3         | 2,067          |
|                      | t2                        | 0,152        | 1,841          |
| calldata             | t3                        | 0,148        | 1,915          |
|                      | t4                        | 0,155        | 1,875          |
|                      | t5                        | 0,15         | 1,912          |
|                      | tamaño                    | 22M          | 12M            |
|                      | tamaño comprimido(tar.gz) | 4,7M         | 12M            |
|                      | tamaño comprimido (gz)    | 4,7M         | 12M            |
|                      | t1                        | 13,9         | 2,165          |
|                      | t2                        | 0,165        | 2,079          |
| calldata_2d          | t3                        | 0,151        | 2,037          |
|                      | t4                        | 0,151        | 2,17           |
|                      | t5                        | 0,154        | 2,043          |
|                      | tamaño                    | 22M          | 11M            |

|  |   |       |       |
|--|---|-------|-------|
|  | tamaño comprimido(tar.gz)                             | 4,7M  | 11M   |
|  | tamaño comprimido (gz)                                | 4,7M  | 11M   |
|  |   |       |       |
|  | suma del tamaño de los 3 ficheros                     | 53,6M | 27,9M |
|  | tamaño de los 3 ficheros comprimidos(tar.gz)          | 12M   | 27M   |
|  | suma del tamaño de los tres ficheros comprimidos (gz) | 12M   | 27M   |

|                      |                           |             |                |
|----------------------|---------------------------|-------------|----------------|
|                      |                           | tomatos     |                |
| tamaño comprimido    | 1,7GB                     | nºmuestras  | 348            |
| tamaño descomprimido | 7,1GB                     | nºSNP       | 868978         |
|                      |                           | vcf2npv (s) | vcfnp2hdf5 (s) |
|                      | t1                        | 4m51        | 19,292         |
|                      | t2                        | 3,115s      | 19,302         |
| variants             | t3                        | 0,183s      | 19.281         |
|                      | t4                        | 0,144s      | 19,391         |
|                      | t5                        | 0,149s      | 19,386         |
|                      | tamaño                    | 200M        | 124M           |
|                      | tamaño comprimido(tar.gz) | 80M         | 123M           |
|                      | tamaño comprimido (gz)    | 81M         | 123M           |
|                      | t1                        | 55m23s      | 45,231         |
|                      | t2                        |             | 1,426          |
| calldata (solo GT)   | t3                        |             | 0,152          |
|                      | t4                        |             | 0,142          |
|                      | t5                        |             | 0,149          |
|                      | tamaño                    | 577M        | 46M            |
|                      | tamaño comprimido(tar.gz) | 23M         | 43M            |

|                       |   |        |        |
|-----------------------|---|--------|--------|
|                       | tamaño comprimido (gz)                                | 23M    | 43M    |
|                       | t1  | 53m56s | 28,458 |
|                       | t2  | 1,476  | 35,977 |
| calldata_2d(sol o GT) | t3  | 0,151  | 22,815 |
|                       | t4  | 0,16   | 19,376 |
|                       | t5  | 0,153  | 18,578 |
|                       | tamaño  | 577M   | 46M    |
|                       | tamaño comprimido(tar.gz)                             | 23M    | 44M    |
|                       | tamaño comprimido (gz)                                | 23M    | 44M    |
|                       |   |        |        |
|                       | suma del tamaño de los 3 ficheros                     | 1354M  | 216M   |
|                       | tamaño de los 3 ficheros comprimidos(tar.gz)          | 125M   | 209M   |
|                       | suma del tamaño de los tres ficheros comprimidos (gz) | 126M   | 209M   |

## VELOCIDADES

|               |      |      |      |      |      |       |       |       |       |      |       |       |       |
|---------------|------|------|------|------|------|-------|-------|-------|-------|------|-------|-------|-------|
|               | 1000 | 5000 | 8000 | 1000 | 1500 | 2000  | 3000  | 3500  | 4000  | 4500 | 5000  | 5500  | 6000  |
| SNP           | 0    | 0    | 0    | 00   | 00   | 00    | 00    | 00    | 00    | 00   | 00    | 00    | 00    |
| t1(s)         | 20   | 36   | 52   | 62   | 87   | 111   | 160   | 188   | 274   | 327  | 541   | 662   | 699   |
| t2(s)         | 17   | 37   | 52   | 62   | 87   | 111   | 160   | 192   | 260   | 303  | 550   | 752   | 689   |
| t3(s)         | 17   | 37   | 53   | 62   | 86   | 111   | 178   | 189   | 256   | 313  | 526   | 726   | 765   |
| t4(s)         | 17   | 36   | 52   | 63   | 86   | 112   | 166   | 191   | 262   | 334  | 551   | 841   | 909   |
| t5(s)         | 17   | 38   | 53   | 62   | 86   | 111   | 157   | 189   | 277   | 313  | 590   | 620   | 837   |
| tmedio<br>(s) | 17,6 | 36,8 | 52,4 | 62,2 | 86,4 | 111,2 | 164,2 | 189,8 | 265,8 | 318  | 551,6 | 720,2 | 779,8 |

|              |        |        |        |        |
|--------------|--------|--------|--------|--------|
| Sin usar del |        |        |        |        |
|              |        |        |        |        |
|              |        |        |        |        |
| SNP          | 100000 | 200000 | 300000 | 400000 |
| t1(s)        | 68     | 114    | 226    | 360    |
| t2(s)        | 61     | 288    | 250    | 421    |
| t3(s)        | 62     | 190    | 246    | 288    |
| t4(s)        | 61     | 142    | 237    | 316    |
| t5(s)        | 61     | 121    | 233    | 314    |
| tmedio(s)    | 62,6   | 171    | 238,4  | 339,8  |

## TIEMPOS INOUT

| Ril                 |           |            |           |            |
|---------------------|-----------|------------|-----------|------------|
|                     | CHUNK=200 | CHUNK=1000 | CHUNK=200 | CHUNK=1000 |
| shuffle<br>fletcher | TRUE      | TRUE       | FALSE     | FALSE      |
| t1(s)               | 8,6       | 8,5        | 8,6       | 8,6        |
| t2(s)               | 8,6       | 8,6        | 8,5       | 8,6        |
| t3(s)               | 8,5       | 8,5        | 8,6       | 8,5        |
| t4(s)               | 8,5       | 8,6        | 8,5       | 8,6        |
| t5(s)               | 8,6       | 8,8        | 8,6       | 8,5        |
| tamaño hdf5         | 517K      | 511K       | 483K      | 485K       |

| Apeki               |                      |            |            |           |            |
|---------------------|----------------------|------------|------------|-----------|------------|
|                     | CHUNK=200            | CHUNK=1000 | CHUNK=2500 | CHUNK=200 | CHUNK=2500 |
| shuffle<br>fletcher | TRUE                 | TRUE       | TRUE       | FALSE     | FALSE      |
| t1(s)               | 357                  | 353        | 369        | 353       | 374        |
| t2(s)               | 361                  | 359        | 364        | 350       | 357        |
| t3(s)               | 346                  | 355        | 367        | 375       | 363        |
| t4(s)               | 348                  | 362        | 368        | 390*      | 359        |
| t5(s)               | 347                  | 357        | 373        | 399*      | 363        |
| tamaño hdf5         | 4.3M                 | 3.4M       | 3.3M       | 4.2M      | 3.4M       |
|                     |                      |            |            |           |            |
|                     | *Ejecutadas a la vez |            |            |           |            |

|         |  |  |  |  |
|---------|--|--|--|--|
| tomatos |  |  |  |  |
|---------|--|--|--|--|

|                    |                           |           |  |                 |
|--------------------|---------------------------|-----------|--|-----------------|
| kept_field=['GT']  |                           |           |  | kept_field=None |
|                    | CHUNK=200                 | CHUNK=200 |  | CHUNK=200       |
| shuffle fletcher y | TRUE                      | FALSE     |  | TRUE            |
| t1(s)              | 1813                      | 1887      |  | 10613           |
| t2(s)              | 1907                      | 1884      |  | 11450*          |
| t3(s)              | 1873                      | 1891      |  | 11053*          |
| t4(s)              | 1886                      | 1872      |  | 11393*          |
| t5(s)              | 1914                      | 2068*     |  | 11229*          |
|                    |                           |           |  |                 |
| tamaño hdf5        | 38M                       | 38M       |  |                 |
|                    |                           |           |  |                 |
|                    |                           |           |  |                 |
|                    | *Con Google Drive abierto |           |  |                 |

## VIDEOS

<https://www.dropbox.com/s/fjw0pjud9edpbue/Videos.zip?dl=0>