(DEMO) Report for ICCS240: DBMS class project: SNPDB

Group Members: Kriangsak T., Hasdin,G. This report refers to this attached repository : SNPBD (https://github.com/Kriangsak1997/DatabaseClassProject)

Please treat this to be a small demo...

First of all, we used docker to run out protgresql database: used commands can be found in the file named "command_involvingDockerandPSQL.txt". In this file, you will find what is used to map data to the container.... since i have no time (more like brain) to build a proper image for it.... Please see the following

In [6]:
```
# filepath  = '/Users/kriangsak1997/Documents/MUIC/Term8/DBMS/Proje
ct/command_involvingDockerandPSQL.txt'
# with open(filepath) as fp:
#    line = fp.readline()
#    cnt = 1
#    while line:
#        print("Line {}: {}".format(cnt, line.strip()))
#        line = fp.readline()
#        cnt += 1
```

Connect to the Database

After having created the database and import the file into it, we will connect to the database using Java. Here with help of **Maven** dependencies, it allows to very easily connect to our database, we can dp so by creating a Maven project. Then access the file: *pom.xml*
and we add the dependencies:

```
</dependency/>

        </groupId/> org.postgresql</groupId/>
         </artifactId/>postgresql</artifactId/>
         </version/>42.2.10</version/>

    </dependency/>
```

What is SNP?

Here, please let me introtduce you with our schema. In case you have not heard of Single Nucleotide Polymorphisms (SNPs).
Single nucleotide polymorphisms, frequently called SNPs (pronounced “snips”), are the most common type of genetic variation among people. Each SNP represents a difference in a single DNA building block, called a nucleotide. For example, a SNP may replace the nucleotide cytosine (C) with the nucleotide thymine (T) in a certain stretch of DNA.

SNPs occur normally throughout a person’s DNA. They occur almost once in every 1,000 nucleotides on average, which means there are roughly 4 to 5 million SNPs in a person's genome. These variations may be unique or occur in many individuals; scientists have found more than 100 million SNPs in populations around the world. Most commonly, these variations are found in the DNA between genes. They can act as biological markers, helping scientists locate genes that are associated with disease. When SNPs occur within a gene or in a regulatory region near a gene, they may play a more direct role in disease by affecting the gene’s function.

Most SNPs have no effect on health or development. Some of these genetic differences, however, have proven to be very important in the study of human health. Researchers have found SNPs that may help predict an individual’s response to certain drugs, susceptibility to environmental factors such as toxins, and risk of developing particular diseases. SNPs can also be used to track the inheritance of disease genes within families. Future studies will work to identify SNPs associated with complex diseases such as heart disease, diabetes, and cancer. source (https://ghr.nlm.nih.gov/primer/genomicresearch/snp)

Schema Detail

We have created the following schema: Note that before inserting the data into our database, we needed to do a bit of porcessing to extract our data out from the origal input data, please refer to (https://github.com/Kriangsak1997/DatabaseClassProject/blob/master/notebooks/processing.ipynb)

SNP_DB(Main_Gene_name varchar, initialAA varchar,finalAA varchar,position_of_Change varchar,Type_of_Variant varchar,dbSNP varchar,Disease_name varchar)

That is,

> *Main_Gene_name* contains the name of the gene for each record, taking *strings* of gene names
>
> *initialAA* contains the amino acid (AA) abbreviation before a change occurs , taking *strings* of AA ,br>
>
> *finalAA* contains the amino acid (AA) abbreviation after a change occurs , taking *strings* of AA
>
> *position_of_Change* contains the position of AA (in the change of AA chains of a gene), taking **string** of position, which later will be altered to integer type
>
> *Type_of_Variant* contains the type of variants for which an SNP causes, taking *strings* of variant types
>
> *dbSNP* take takes in database code used to access more information regarding a change in NCBI SNP database, taking strings of DB-code
>
> *Disease_name* contains the name of diseases, if applicable, caused by SNPs, taking *strings* of disease names.

Abbreviations and Full names

The following dataframe displays to you the full AA names corresponding to abbreviations used in our database

In [7]:
```
AminoAcid_name =pd.read_csv('/Users/kriangsak1997/Documents/MUIC/Term8/DBMS/Project/aanames.csv')
AminoAcid_name
```

Out[7]:

	Abbreviation	Ful name
0	Ala	Alanine
1	Arg	Arginine
2	Asn	Asparagine
3	Asp	Aspartic acid (Aspartate)
4	Cys	Cysteine
5	Gln	Glutamine
6	Glu	Glutamic acid (Glutamate)
7	Gly	Glycine
8	His	Histidine
9	Ile	Isoleucine
10	Leu	Leucine
11	Lys	Lysine
12	Met	Methionine
13	Phe	Phenylalanine
14	Pro	Proline
15	Ser	Serine
16	Thr	Threonine
17	Trp	Tryptophan
18	Tyr	Tyrosine
19	Val	Valine

Now we can query from out database for all: CRUD queries

From this, we take a data-analytic approach to our database. That is, we will apply a small mining algorithm on the given dataset.

Associative Rule Mining with Apriori Algorithm

for those wishing to understand more about this concept, consult (https://blog.usejournal.com/association-rule-mining-apriori-algorithm-c517f8d7c54c) There are a few things to introduce for those new to Associative Rule Mining or, perhaps also, Apriori Algorithm.
the results of out experiments is in the file: notebooks/Association Rule Mining with Apriori algorithm.ipynb with a few tials of different hyperparemeters (supports, Confidence). It turns out that the with conditional probabiity of 25%, this gives out 3 frequent item set(of 2 elements):

('Asp', 'Asn'), ('Tyr', 'Cys'), ('Glu', 'Lys')

This will be examined later in this demo.

In addition to the first submission, there is a need for a moer thorough anaysis of the data. To do so, the problem that was not fixed, purposely actually ^^, is that we need to convert the type of attibute **position_of_change** into an **integer** type. It is very simple to do so, we can simply run the following command to our datbase.

```
ALTER TABLE snp_db ALTER COLUMN position_of_change TYPE INT USING
position_of_change::integer;
```

From here on, we can do analysis associated with position of chnages and our obtained frequent itemsets obtained from associative rule mining. For simplicity, let's bring in a few functions which will later be used in this demo.

In [6]:
```python
import psycopg2
import pandas as pd
import numpy as np
from apyori import apriori
import matplotlib.pyplot as plt
from sqlalchemy import create_engine
"""
This connection function will acts as a connection factory as it is
in our ConnectionFactory.java
in this repository.
"""
def connection(user,pwd,host,port,db):
    try:
       connection = psycopg2.connect(user=user,
                                    password=pwd,
                                    host=host,
                                    port=port,
                                    database=db)
       cursor = connection.cursor()
       return cursor
    except (Exception, psycopg2.Error) as error :
        print ("Error while fetching data from PostgreSQL", error)

"""
this select class will return the output from input queries and ret
urn, for the moment, as a numpy array object.
"""
user ="postgres"
pwd = "1997"
host ="localhost"
port = "5432"
db= "postgres"
def select(q):
    c = connection(user,pwd,host,port,db)
    c.execute(q)
    arr = np.array(c.fetchall())
    return arr
```

In [51]:
```
snp =pd.DataFrame(select("select initialAA,finalAA from snp_db"))
snp.columns=[ 'initialAA', 'finalAA']
snps = []
for row in range(0,len(snp)):
    snps.append([str(snp.values[row,j]) for j in range(0,2)])
association_rules = apriori(snps,min_support=0.0093,min_confidence=
0.250,min_lift =3,min_length=2)
association_results =list(association_rules)
finals = []
rows = []
for item in association_results:
#     to display the rule, the support, the confidence, and lift fo
r each rule in a more clear way:

    # first index of the inner list
    # Contains base item and add item
    eachrow=[]
    pair = item[0]
    items = [x for x in pair]
#     print("Rule: " + items[0] + " -> " + items[1])
    eachrow.append(items[0] + " -> " + items[1])

    #second index of the inner list
#     print("Support: " + str(item[1]))
    eachrow.append(item[1])

    #third index of the list located at 0th
    #of the third index of the inner list

#     print("Confidence: " + str(item[2][0][2]))
    eachrow.append(item[2][0][2])
#     print("Lift: " + str(item[2][0][3]))
    eachrow.append(item[2][0][3])
    rows.append(eachrow)
#     print("=====================================")
miningresults = pd.DataFrame(rows)
cols = ["rule", "Support", "Confidence", "Lift"]
miningresults.columns = cols
```

In [55]:
```
miningresults
# miningresults.to_csv(r'/Users/kriangsak1997/Documents/MUIC/Term8/
DBMS/Project/miningResults.csv', index = False)
```

Out[55]:

	rule	Support	Confidence	Lift
0	Asn -> Asp	0.020755	0.283619	3.397908
1	Tyr -> Cys	0.021931	0.254751	4.943691
2	Lys -> Glu	0.031555	0.369445	5.063446

Note that we have added the result into the 8th case of our switch case class so that the user can see this obtained result

What can see learn from the following rules

we shall try to zoom in into each of an associative frequent itemsets and see what can be drawn from it. For example, we can ask the following questions.

> (1) What type of variants are caused by the obtained rules.
> (2) What are are the positions of change when we specify with obtained initial and final AAs. we can bin this, say, into 3 subintervals. From this quetion, we can deepen the quetion into: once binneed into 3 subintervals, which subinterval containes the largest number of changes recorded

(1) What type of variants are caused by the obtained rules

In [8]:
```
#Prepare the queries for the 3 rules
final_Queries = ["select count(main_gene_name) from snp_db where in
itialaa='Asp' and finalaa = 'Asn' group by type_of_variant;",
                "select count(main_gene_name) from snp_db where ini
tialaa='Tyr' and finalaa = 'Cys' group by type_of_variant; ",
                 "select count(main_gene_name) from snp_db where in
itialaa='Glu' and finalaa = 'Lys' group by type_of_variant;"]
records= [select(final_Queries[i]).T.flatten().tolist() for i in ra
nge(3)]
framessss = pd.DataFrame(records)
framessss.insert(0,"Rule",["Asp -> Asn", "Tyr -> Cys", "Glu -> Lys"
],True)
framessss.columns = ["Rule\Type of Variant","Disease","Polymorphism
","Unclassified"]
#note that pqsl return the count in alphabetical order.
framessss.set_index(['Rule\Type of Variant'])
```

Out[8]:

	Disease	Polymorphism	Unclassified
Rule\Type of Variant			
Asp -> Asn	457	592	147
Tyr -> Cys	574	285	80
Glu -> Lys	785	813	214

In [9]:
```
# framessss.to_csv(r'/Users/kriangsak1997/Documents/MUIC/Term8/DBMS
/Project/frequent_thingy.csv', index = False)
```

In [10]:
```
# check = "select * from frequent_things"
# select(check)
```

Here we go, the above dataframe displays different types of variants based on the given associative rules. It is obvious to see that 2 out of 3 rules (Asp -> Asn,Glu -> Lys), when these changes occur, they give rise to polymorphism more than the other two types of genetic variations

(2) Binning of positions of changes

like we said earilier, we would like to know What are are the positions of change when we specify with obtained initial and final AAs. we can bin this, say, into 3 subintervals. From this quetion, we can deepen the quetion into: once binneed into 3 subintervals, which subinterval containes the largest number of changes recorded. Here is the plans of what we are going to do in the next few cells

> First, we want to bin the entire dataset
> Second, we will do the same for each of the 3 obtained assiciative frequent itemset.
> finally, we will compare the result accordingly.

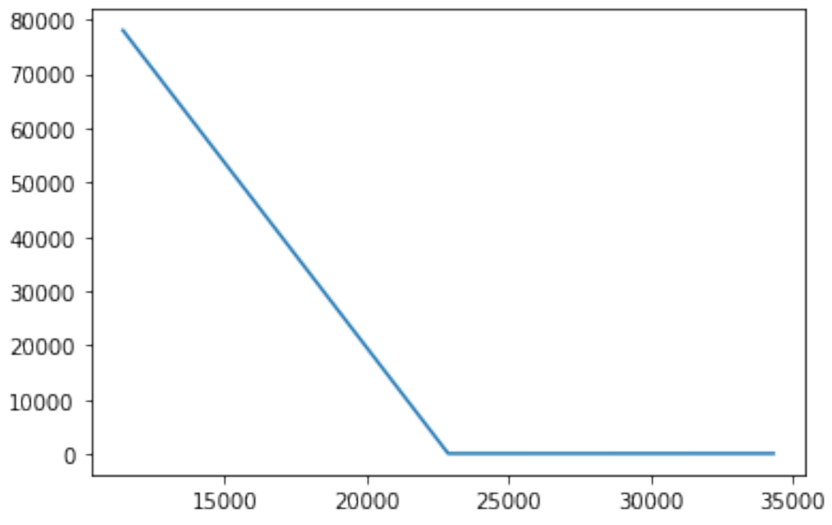
Binning the whole dataset

In [11]:
```
count_main, bin_main = np.histogram(select("select position_of_chan
ge from snp_db"),3)
# plt.hist(bins[:-1], bins, weights=counts)
```

In [12]:
```
intv= [11439.6, 22877.3,34315.0]
vals = count_main.flatten().tolist()
vals
```

Out[12]:
```
[78023, 108, 114]
```

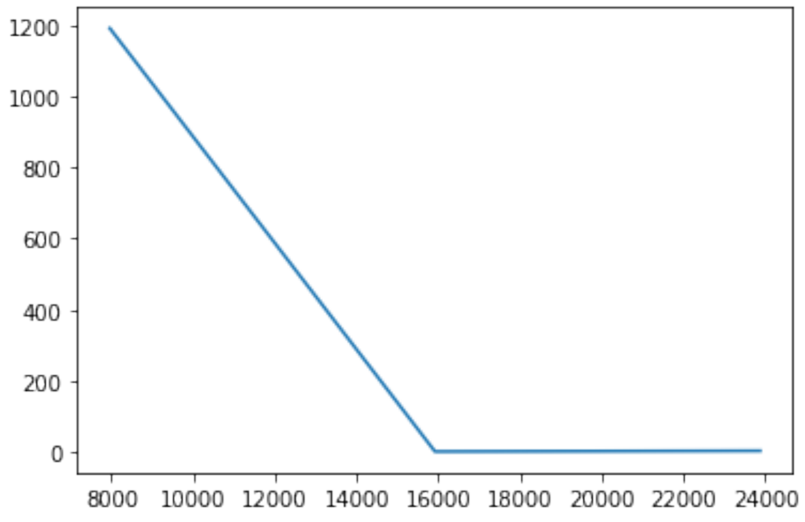
```
In [13]: plot_main = plt.plot(intv,vals)
```



Binning the case when Asp changes into Asn

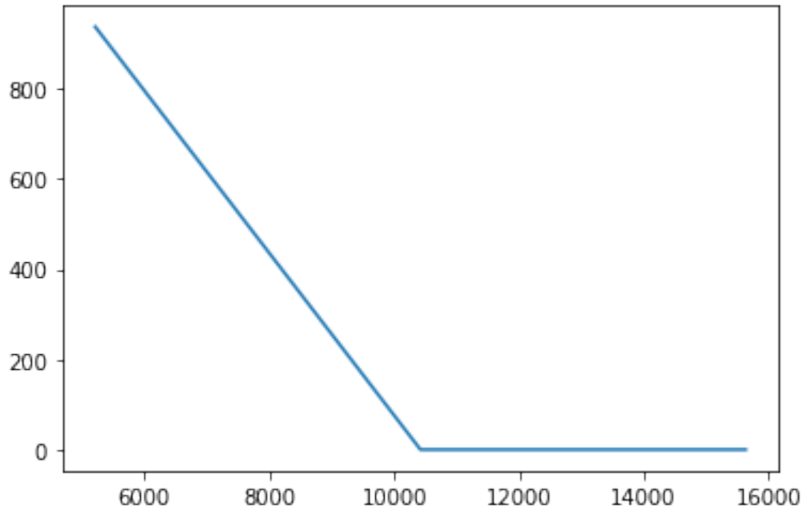
```
In [14]: count_1, bins_1 = np.histogram(select("select position_of_change fr
         om snp_db where initialaa='Asp' and finalaa = 'Asn'"),3)
         plt.plot(bins_1.flatten().tolist()[1:],count_1.flatten().tolist())
         count_1,bins_1
```

```
Out[14]: (array([1192,    1,    3]),
          array([2.00000000e+00, 7.95866667e+03, 1.59153333e+04, 2.38720000
         e+04]))
```



Binning the case when Tyr changes into Cys

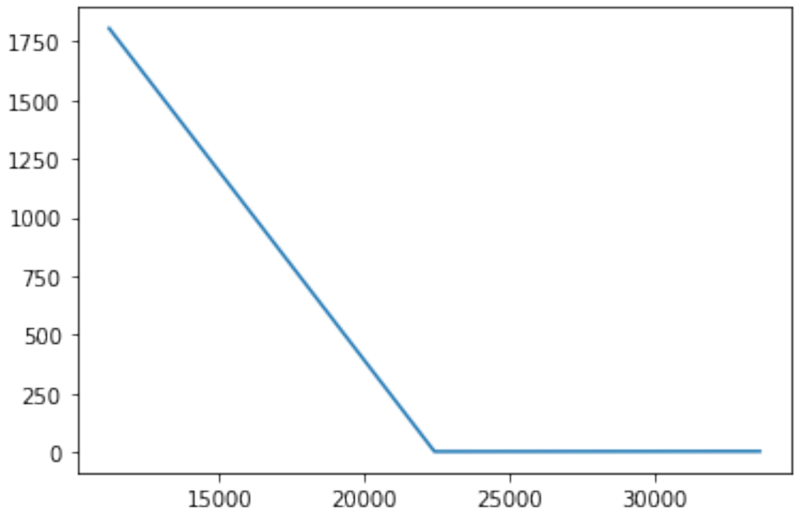
In [59]:
```
count_2, bins_2 = np.histogram(select("select position_of_change from snp_db where initialaa='Tyr' and finalaa = 'Cys'"),3)
plt.plot(bins_2.flatten().tolist()[1:],count_2.flatten().tolist())
count_2,bins_2
from IPython.display import Image
```



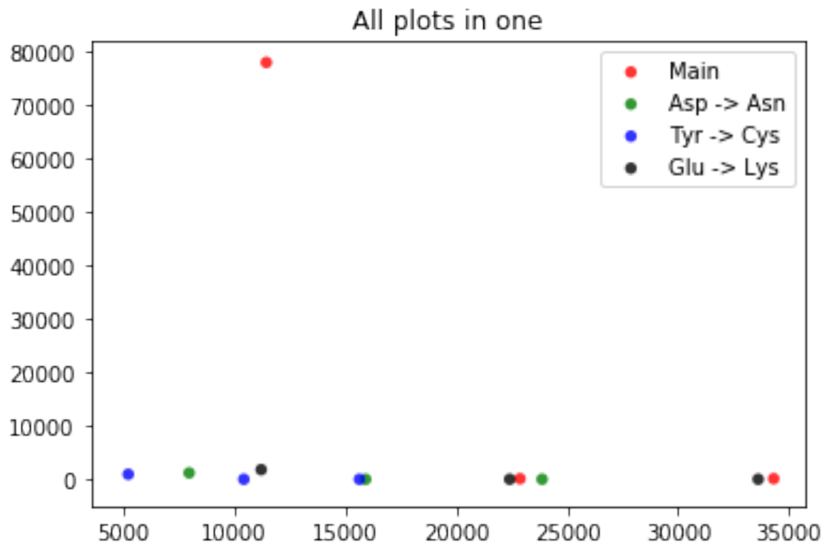
Binning the case when Glu changes into Lys

In [16]:
```
count_3, bins_3 = np.histogram(select("select position_of_change from snp_db where initialaa='Glu' and finalaa = 'Lys'"),3)
plt3 =plt.plot(bins_3.flatten().tolist()[1:],count_3.flatten().tolist())
count_3,bins_3
```

Out[16]:
```
(array([1805,    3,    4]),
 array([2.00000000e+00, 1.12066667e+04, 2.24113333e+04, 3.36160000
e+04]))
```



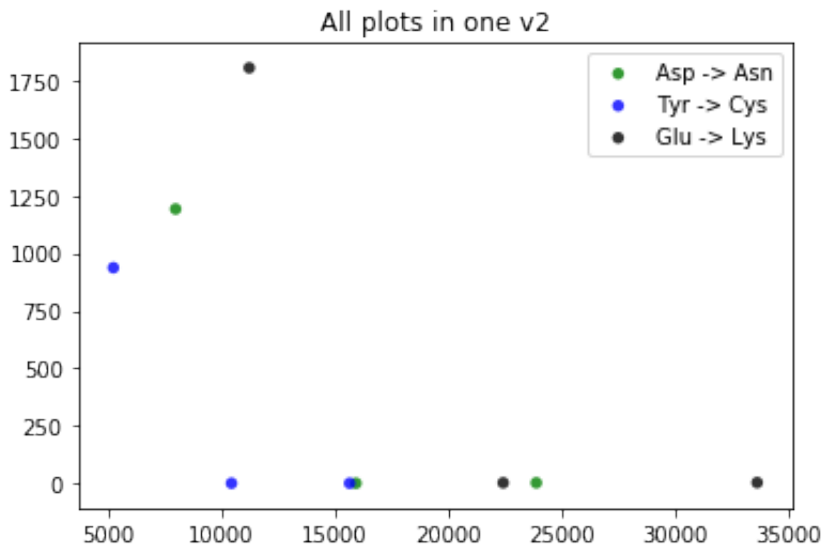
In [17]:
```
colors = (0,0,0)
area = np.pi*3

# Plot

g1 = (bin_main.flatten().tolist()[1:], count_main.flatten().tolist(
))
g2 = (bins_1.flatten().tolist()[1:], count_1.flatten().tolist())
g3 = (bins_2.flatten().tolist()[1:], count_2.flatten().tolist())
g4 = (bins_3.flatten().tolist()[1:], count_3.flatten().tolist())
data = (g1, g2, g3,g4)
colors = ("red", "green", "blue","black")
groups = (["Main","Asp -> Asn", "Tyr -> Cys", "Glu -> Lys"])

# Create plot
fig = plt.figure()
ax = fig.add_subplot(1, 1, 1)

for data, color, group in zip(data, colors, groups):
    x, y = data
    ax.scatter(x, y, alpha=0.8, c=color, edgecolors='none', s=30, l
abel=group)

plt.title('All plots in one')
plt.legend(loc=0)
plt.show()
```



Plotting with the main dataset makes it hard to see the actual trend for the subplots: all the frequent iremsets.

We will try to construct a new plot excluding the main dataset

In [18]:
```
colors = (0,0,0)
area = np.pi*3

# g1 = (bin_main.flatten().tolist()[1:], count_main.flatten().tolis
t())
g2 = (bins_1.flatten().tolist()[1:], count_1.flatten().tolist())
g3 = (bins_2.flatten().tolist()[1:], count_2.flatten().tolist())
g4 = (bins_3.flatten().tolist()[1:], count_3.flatten().tolist())
data = ( g2, g3,g4)
colors = ( "green", "blue","black")
groups = (["Asp -> Asn", "Tyr -> Cys", "Glu -> Lys"])

# Create plot
fig = plt.figure()
ax = fig.add_subplot(1, 1, 1)

for data, color, group in zip(data, colors, groups):
    x, y = data
    ax.scatter(x, y, alpha=0.8, c=color, edgecolors='none', s=30, l
abel=group)

plt.title('All plots in one v2')
plt.legend(loc=0)
plt.show()
```



from here we can see that the the most significant intervals are the first interval of every subplot.

In addition to that, we can also try to check for which of the frequent itemset, which one has the most number of changes cross all subitervals

In [57]:
```
countsss = [count_1, count_2, count_3]
sumcol = [np.sum(count_1),np.sum(count_2),np.sum(count_3)]
countframes = pd.DataFrame(countsss)
countframes.columns= ["1st-inv'l","2nd-inv'l","3rd-inv'l"]
countframes
countframes.insert(0,"Rule",["Asp -> Asn", "Tyr -> Cys", "Glu -> Ly
s"],True)
countframes.insert(4,"total",sumcol,True)
countframes.columns = ["Rule\intvl","1st-inv'l","2nd-inv'l","3rd-in
v'l","total"]
#note that pqsl return the count in alphabetical order.
countframes.set_index(['Rule\intvl'])
new = countframes
new
```

Out[57]:

	Rule\intvl	1st-inv'l	2nd-inv'l	3rd-inv'l	total
0	Asp -> Asn	1192	1	3	1196
1	Tyr -> Cys	937	1	1	939
2	Glu -> Lys	1805	3	4	1812

As we can see from the summary table, when Asp changes into Asn, it has the largest number of changes. In fact, we also know this from the outout from our associative mining. That is, we can pinpoint this fact by looking at the conditional probabilities, confidence values.

In [56]:
```
# new.to_csv(r'/Users/kriangsak1997/Documents/MUIC/Term8/DBMS/Proje
ct/count_per_interval.csv', index = False)
```

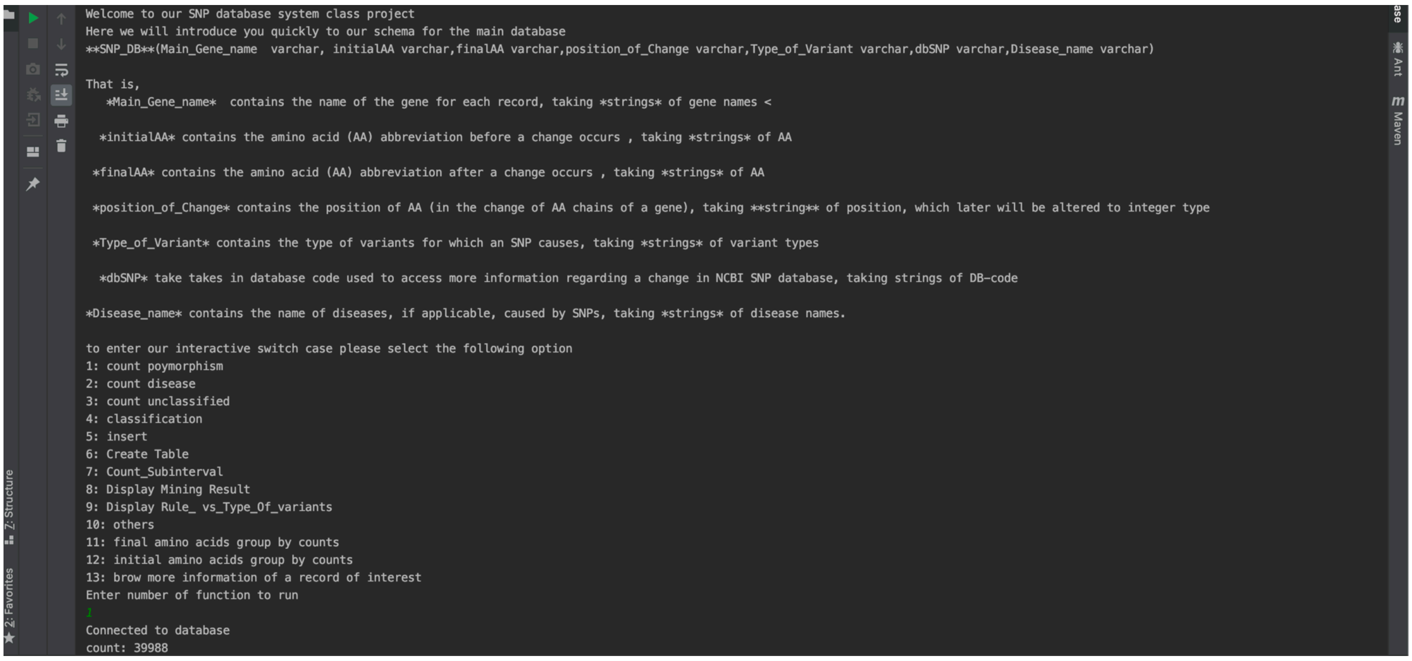
Put the rules aside

Apart from the ontained results, we can also pose a few quetions directly to our databse. Let's assume that there exists a curious user wanting to know the followig:

- what is the count the each amino acid in the database: initial and final counts: to see this please select case 11 ans 13.
- what if the user wants to know more about a specific record, then go ahead with case 13: where the input parameter is taken from attribute "dbSNP". The user shall have a brower opening the NBCI page linked to the specified record. Let's see how each of the switch cases works!!

Case 1: Count polymorphism

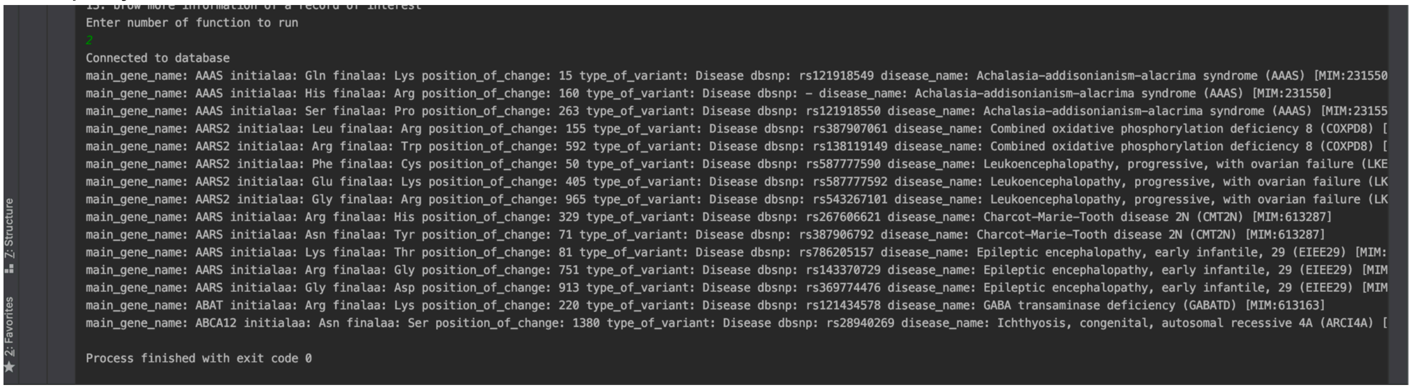
this query return to the user: the total number of polymorphism cases in the database



```
Welcome to our SNP database system class project
Here we will introduce you quickly to our schema for the main database
**SNP_DB**(Main_Gene_name  varchar, initialAA varchar,finalAA varchar,position_of_Change varchar,Type_of_Variant varchar,dbSNP varchar,Disease_name varchar)

That is,
   *Main_Gene_name*  contains the name of the gene for each record, taking *strings* of gene names <

  *initialAA* contains the amino acid (AA) abbreviation before a change occurs , taking *strings* of AA

 *finalAA* contains the amino acid (AA) abbreviation after a change occurs , taking *strings* of AA

 *position_of_Change* contains the position of AA (in the change of AA chains of a gene), taking **string** of position, which later will be altered to integer type

 *Type_of_Variant* contains the type of variants for which an SNP causes, taking *strings* of variant types

  *dbSNP* take takes in database code used to access more information regarding a change in NCBI SNP database, taking strings of DB-code

*Disease_name* contains the name of diseases, if applicable, caused by SNPs, taking *strings* of disease names.

to enter our interactive switch case please select the following option
1: count poymorphism
2: count disease
3: count unclassified
4: classification
5: insert
6: Create Table
7: Count_Subinterval
8: Display Mining Result
9: Display Rule_ vs_Type_Of_variants
10: others
11: final amino acids group by counts
12: initial amino acids group by counts
13: brow more information of a record of interest
Enter number of function to run
1
Connected to database
count: 39988
```

Case 2: Count disease

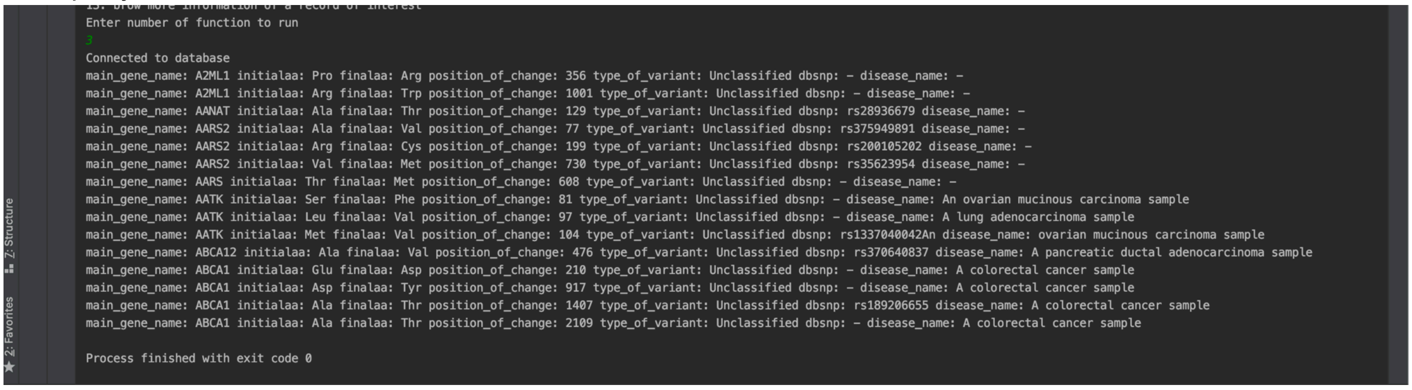
this query return to the user: the total number of disease cases in the database



```
Enter number of function to run
2
Connected to database
main_gene_name: AAAS initialaa: Gln finalaa: Lys position_of_change: 15 type_of_variant: Disease dbsnp: rs121918549 disease_name: Achalasia-addisonianism-alacrima syndrome (AAAS) [MIM:231550
main_gene_name: AAAS initialaa: His finalaa: Arg position_of_change: 160 type_of_variant: Disease dbsnp: - disease_name: Achalasia-addisonianism-alacrima syndrome (AAAS) [MIM:231550]
main_gene_name: AAAS initialaa: Ser finalaa: Pro position_of_change: 263 type_of_variant: Disease dbsnp: rs121918550 disease_name: Achalasia-addisonianism-alacrima syndrome (AAAS) [MIM:23155
main_gene_name: AARS2 initialaa: Leu finalaa: Arg position_of_change: 155 type_of_variant: Disease dbsnp: rs387907061 disease_name: Combined oxidative phosphorylation deficiency 8 (COXPD8) [
main_gene_name: AARS2 initialaa: Arg finalaa: Trp position_of_change: 592 type_of_variant: Disease dbsnp: rs138119149 disease_name: Combined oxidative phosphorylation deficiency 8 (COXPD8) [
main_gene_name: AARS2 initialaa: Phe finalaa: Cys position_of_change: 50 type_of_variant: Disease dbsnp: rs587777590 disease_name: Leukoencephalopathy, progressive, with ovarian failure (LKE
main_gene_name: AARS2 initialaa: Glu finalaa: Lys position_of_change: 405 type_of_variant: Disease dbsnp: rs587777592 disease_name: Leukoencephalopathy, progressive, with ovarian failure (LK
main_gene_name: AARS2 initialaa: Gly finalaa: Arg position_of_change: 965 type_of_variant: Disease dbsnp: rs543267101 disease_name: Leukoencephalopathy, progressive, with ovarian failure (LK
main_gene_name: AARS initialaa: Arg finalaa: His position_of_change: 329 type_of_variant: Disease dbsnp: rs267606621 disease_name: Charcot-Marie-Tooth disease 2N (CMT2N) [MIM:613287]
main_gene_name: AARS initialaa: Asn finalaa: Tyr position_of_change: 71 type_of_variant: Disease dbsnp: rs387906792 disease_name: Charcot-Marie-Tooth disease 2N (CMT2N) [MIM:613287]
main_gene_name: AARS initialaa: Lys finalaa: Thr position_of_change: 81 type_of_variant: Disease dbsnp: rs786205157 disease_name: Epileptic encephalopathy, early infantile, 29 (EIEE29) [MIM:
main_gene_name: AARS initialaa: Arg finalaa: Gly position_of_change: 751 type_of_variant: Disease dbsnp: rs143370729 disease_name: Epileptic encephalopathy, early infantile, 29 (EIEE29) [MIM
main_gene_name: AARS initialaa: Gly finalaa: Asp position_of_change: 913 type_of_variant: Disease dbsnp: rs369774476 disease_name: Epileptic encephalopathy, early infantile, 29 (EIEE29) [MIM
main_gene_name: ABAT initialaa: Arg finalaa: Lys position_of_change: 220 type_of_variant: Disease dbsnp: rs121434578 disease_name: GABA transaminase deficiency (GABATD) [MIM:613163]
main_gene_name: ABCA12 initialaa: Asn finalaa: Ser position_of_change: 1380 type_of_variant: Disease dbsnp: rs28940269 disease_name: Ichthyosis, congenital, autosomal recessive 4A (ARCI4A) [

Process finished with exit code 0
```

Case 3: Count unclassified

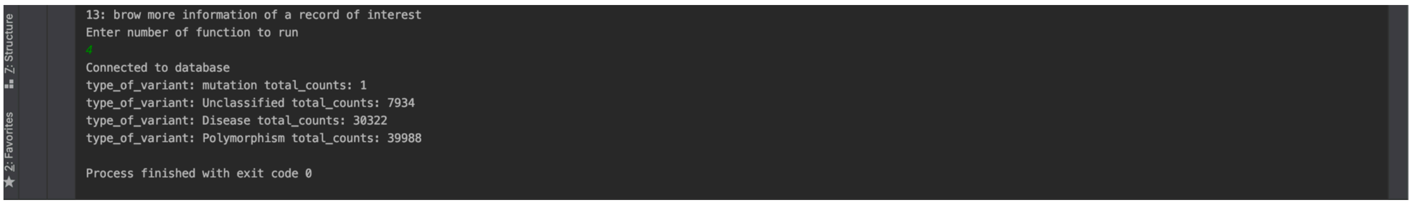
this query return to the user: the total number of unclassified cases in the database



```
Enter number of function to run
3
Connected to database
main_gene_name: A2ML1 initialaa: Pro finalaa: Arg position_of_change: 356 type_of_variant: Unclassified dbsnp: - disease_name: -
main_gene_name: A2ML1 initialaa: Arg finalaa: Trp position_of_change: 1001 type_of_variant: Unclassified dbsnp: - disease_name: -
main_gene_name: AANAT initialaa: Ala finalaa: Thr position_of_change: 129 type_of_variant: Unclassified dbsnp: rs28936679 disease_name: -
main_gene_name: AARS2 initialaa: Ala finalaa: Val position_of_change: 77 type_of_variant: Unclassified dbsnp: rs375949891 disease_name: -
main_gene_name: AARS2 initialaa: Arg finalaa: Cys position_of_change: 199 type_of_variant: Unclassified dbsnp: rs200105202 disease_name: -
main_gene_name: AARS2 initialaa: Val finalaa: Met position_of_change: 730 type_of_variant: Unclassified dbsnp: rs35623954 disease_name: -
main_gene_name: AARS initialaa: Thr finalaa: Met position_of_change: 608 type_of_variant: Unclassified dbsnp: - disease_name: -
main_gene_name: AATK initialaa: Ser finalaa: Phe position_of_change: 81 type_of_variant: Unclassified dbsnp: - disease_name: An ovarian mucinous carcinoma sample
main_gene_name: AATK initialaa: Leu finalaa: Val position_of_change: 97 type_of_variant: Unclassified dbsnp: - disease_name: A lung adenocarcinoma sample
main_gene_name: AATK initialaa: Met finalaa: Val position_of_change: 104 type_of_variant: Unclassified dbsnp: rs1337040042An disease_name: ovarian mucinous carcinoma sample
main_gene_name: ABCA12 initialaa: Ala finalaa: Val position_of_change: 476 type_of_variant: Unclassified dbsnp: rs370640837 disease_name: A pancreatic ductal adenocarcinoma sample
main_gene_name: ABCA1 initialaa: Glu finalaa: Asp position_of_change: 210 type_of_variant: Unclassified dbsnp: - disease_name: A colorectal cancer sample
main_gene_name: ABCA1 initialaa: Asp finalaa: Tyr position_of_change: 917 type_of_variant: Unclassified dbsnp: - disease_name: A colorectal cancer sample
main_gene_name: ABCA1 initialaa: Ala finalaa: Thr position_of_change: 1407 type_of_variant: Unclassified dbsnp: rs189206655 disease_name: A colorectal cancer sample
main_gene_name: ABCA1 initialaa: Ala finalaa: Thr position_of_change: 2109 type_of_variant: Unclassified dbsnp: - disease_name: A colorectal cancer sample

Process finished with exit code 0
```

Case 4: Classification,

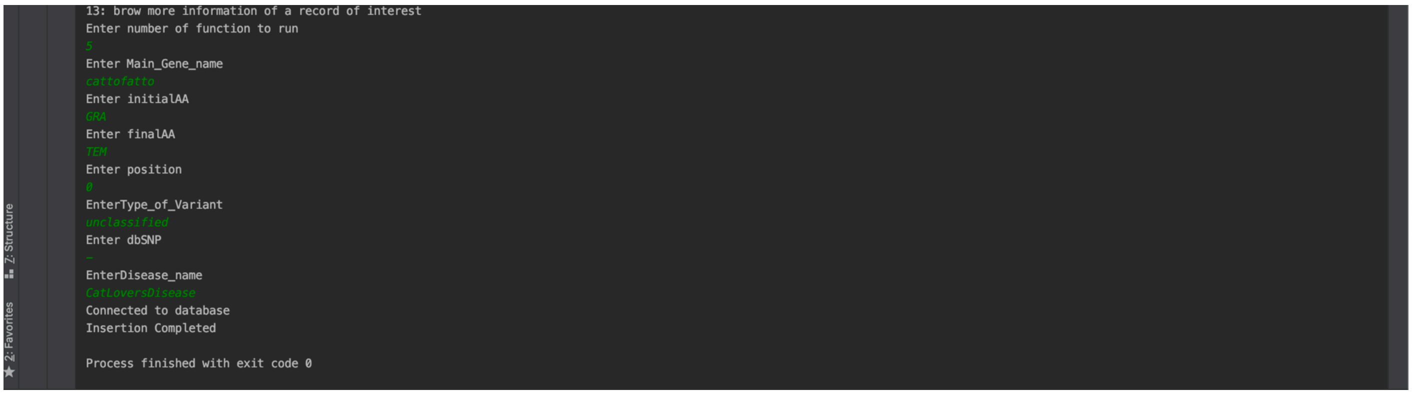
I know this is just the aggregation of case 1,2 and 3, but why not, let's just see it!



```
13: brow more information of a record of interest
Enter number of function to run
4
Connected to database
type_of_variant: mutation total_counts: 1
type_of_variant: Unclassified total_counts: 7934
type_of_variant: Disease total_counts: 30322
type_of_variant: Polymorphism total_counts: 39988

Process finished with exit code 0
```

Case 5: Insert record

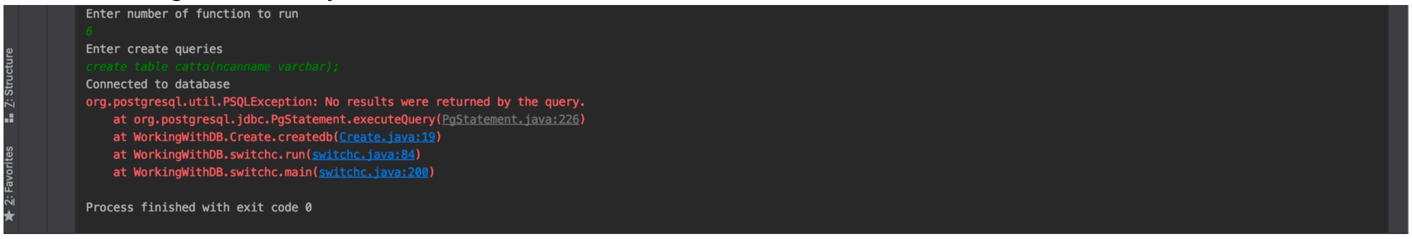
This case takes care of record insertion



```
13: brow more information of a record of interest
Enter number of function to run
5
Enter Main_Gene_name
cattofatto
Enter initialAA
GRA
Enter finalAA
TEM
Enter position
0
EnterType_of_Variant
unclassified
Enter dbSNP
-
EnterDisease_name
CatLoversDisease
Connected to database
Insertion Completed

Process finished with exit code 0
```

Case 6: Create Table

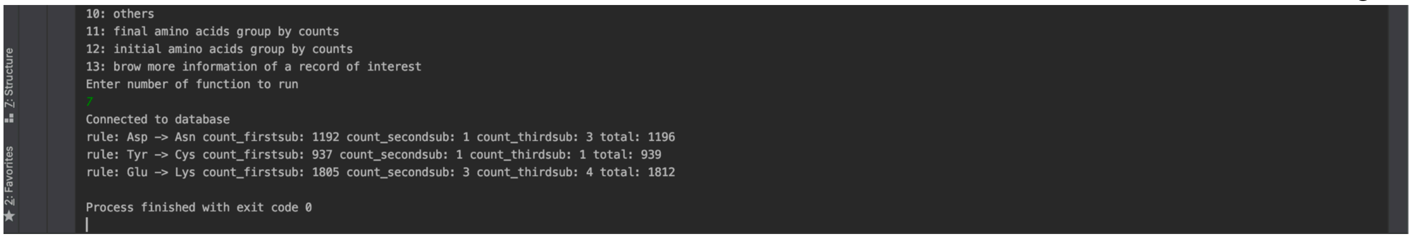
This case takes care of creating a new table...it shows some sort of error, but it works.... I dont know how to fix this bug...admitedly.



```
Enter number of function to run
6
Enter create queries
create table catto(ncanname varchar);
Connected to database
org.postgresql.util.PSQLException: No results were returned by the query.
    at org.postgresql.jdbc.PgStatement.executeQuery(PgStatement.java:226)
    at WorkingWithDB.Create.createdb(Create.java:19)
    at WorkingWithDB.switchc.run(switchc.java:84)
    at WorkingWithDB.switchc.main(switchc.java:200)

Process finished with exit code 0
```

Case 7: Count_Subinterval

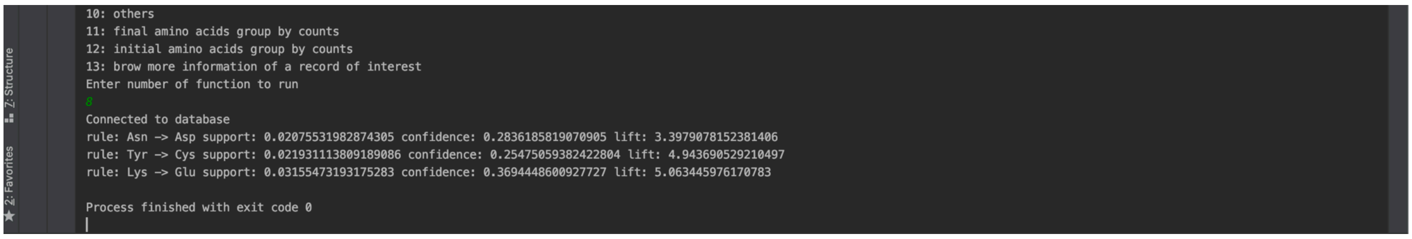
This case returns to the user the counts for each subintervals resulted from our Associative rule mining



```
10: others
11: final amino acids group by counts
12: initial amino acids group by counts
13: brow more information of a record of interest
Enter number of function to run
7
Connected to database
rule: Asp -> Asn count_firstsub: 1192 count_secondsub: 1 count_thirdsub: 3 total: 1196
rule: Tyr -> Cys count_firstsub: 937 count_secondsub: 1 count_thirdsub: 1 total: 939
rule: Glu -> Lys count_firstsub: 1805 count_secondsub: 3 count_thirdsub: 4 total: 1812

Process finished with exit code 0
```

Case 8: Display Mining Result

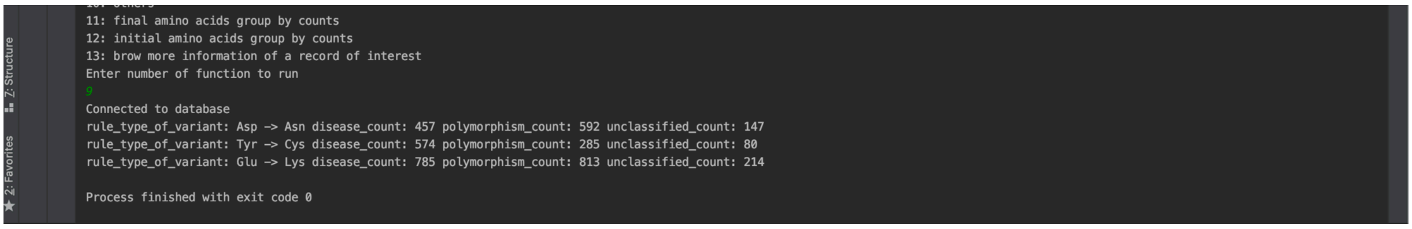
This case returns to you the result of our mining on the database: and yes I know it shuold have come before case 7....but whatever.



```
10: others
11: final amino acids group by counts
12: initial amino acids group by counts
13: brow more information of a record of interest
Enter number of function to run
8
Connected to database
rule: Asn -> Asp support: 0.02075531982874305 confidence: 0.2836185819070905 lift: 3.3979078152381406
rule: Tyr -> Cys support: 0.021931113809189086 confidence: 0.25475059382422804 lift: 4.943690529210497
rule: Lys -> Glu support: 0.03155473193175283 confidence: 0.3694448600927727 lift: 5.063445976170783

Process finished with exit code 0
```

Case 9: Display Rule_ vs_Type_Of_variants

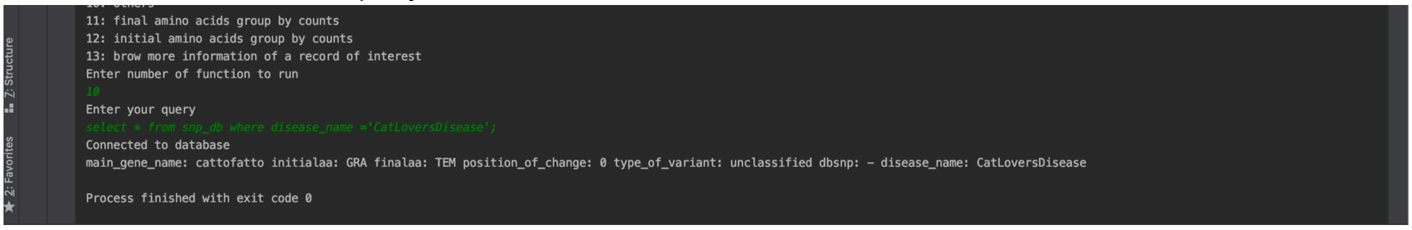
This case presents to the user, with three obtained associative frequent itemsets, the catagorizations of variants associated to each rule.



```
11: final amino acids group by counts
12: initial amino acids group by counts
13: brow more information of a record of interest
Enter number of function to run
9
Connected to database
rule_type_of_variant: Asp -> Asn disease_count: 457 polymorphism_count: 592 unclassified_count: 147
rule_type_of_variant: Tyr -> Cys disease_count: 574 polymorphism_count: 285 unclassified_count: 80
rule_type_of_variant: Glu -> Lys disease_count: 785 polymorphism_count: 813 unclassified_count: 214

Process finished with exit code 0
```

Case 10: others

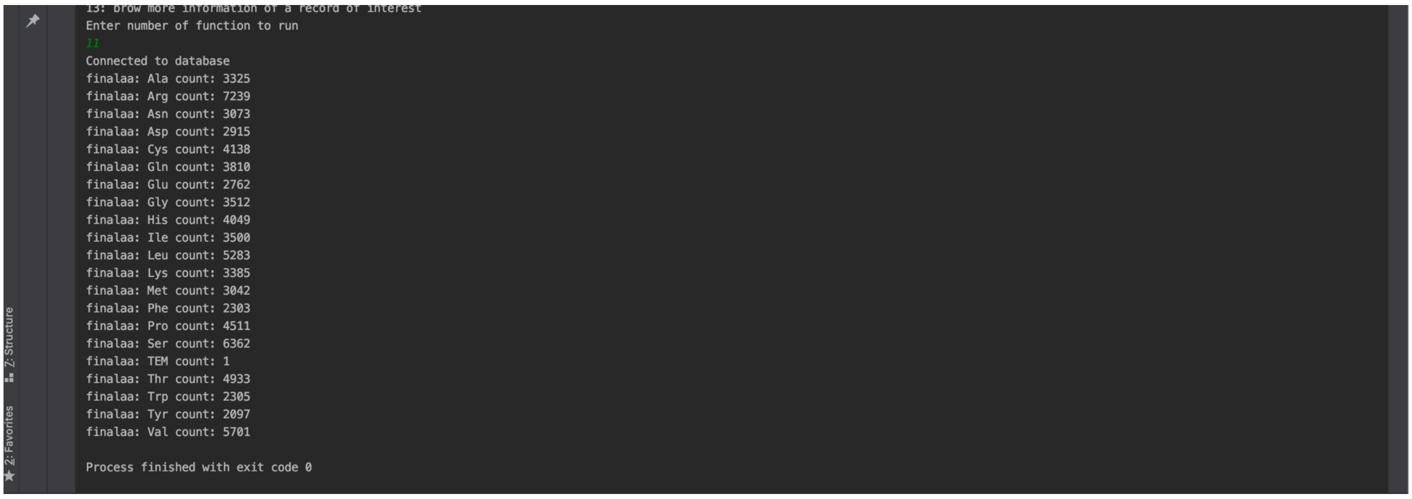
This case allows users to query from the database



```
11: final amino acids group by counts
12: initial amino acids group by counts
13: brow more information of a record of interest
Enter number of function to run
10
Enter your query
select * from snp_db where disease_name ='CatLoversDisease';
Connected to database
main_gene_name: cattofatto initialaa: GRA finalaa: TEM position_of_change: 0 type_of_variant: unclassified dbsnp: - disease_name: CatLoversDisease

Process finished with exit code 0
```

Case 11: final amino acids group by counts

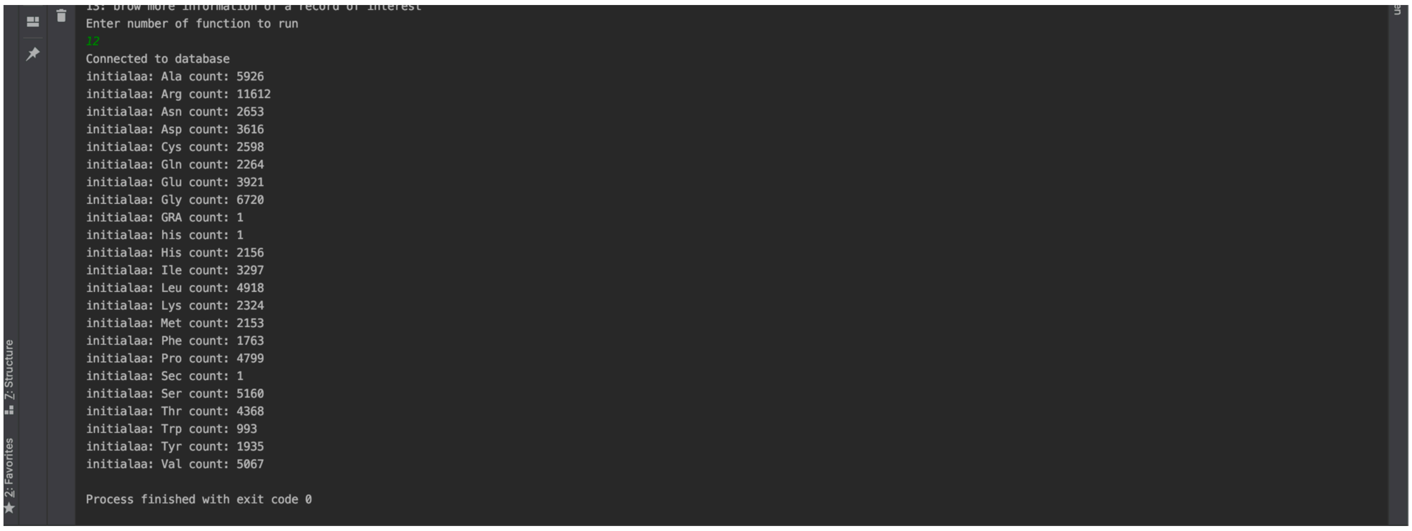
This case counts the final amino acids



```
13: brow more information of a record of interest
Enter number of function to run
11
Connected to database
finalaa: Ala count: 3325
finalaa: Arg count: 7239
finalaa: Asn count: 3073
finalaa: Asp count: 2915
finalaa: Cys count: 4138
finalaa: Gln count: 3810
finalaa: Glu count: 2762
finalaa: Gly count: 3512
finalaa: His count: 4049
finalaa: Ile count: 3500
finalaa: Leu count: 5283
finalaa: Lys count: 3385
finalaa: Met count: 3042
finalaa: Phe count: 2303
finalaa: Pro count: 4511
finalaa: Ser count: 6362
finalaa: TEM count: 1
finalaa: Thr count: 4933
finalaa: Trp count: 2305
finalaa: Tyr count: 2097
finalaa: Val count: 5701

Process finished with exit code 0
```

Case 12: initial amino acids group by count

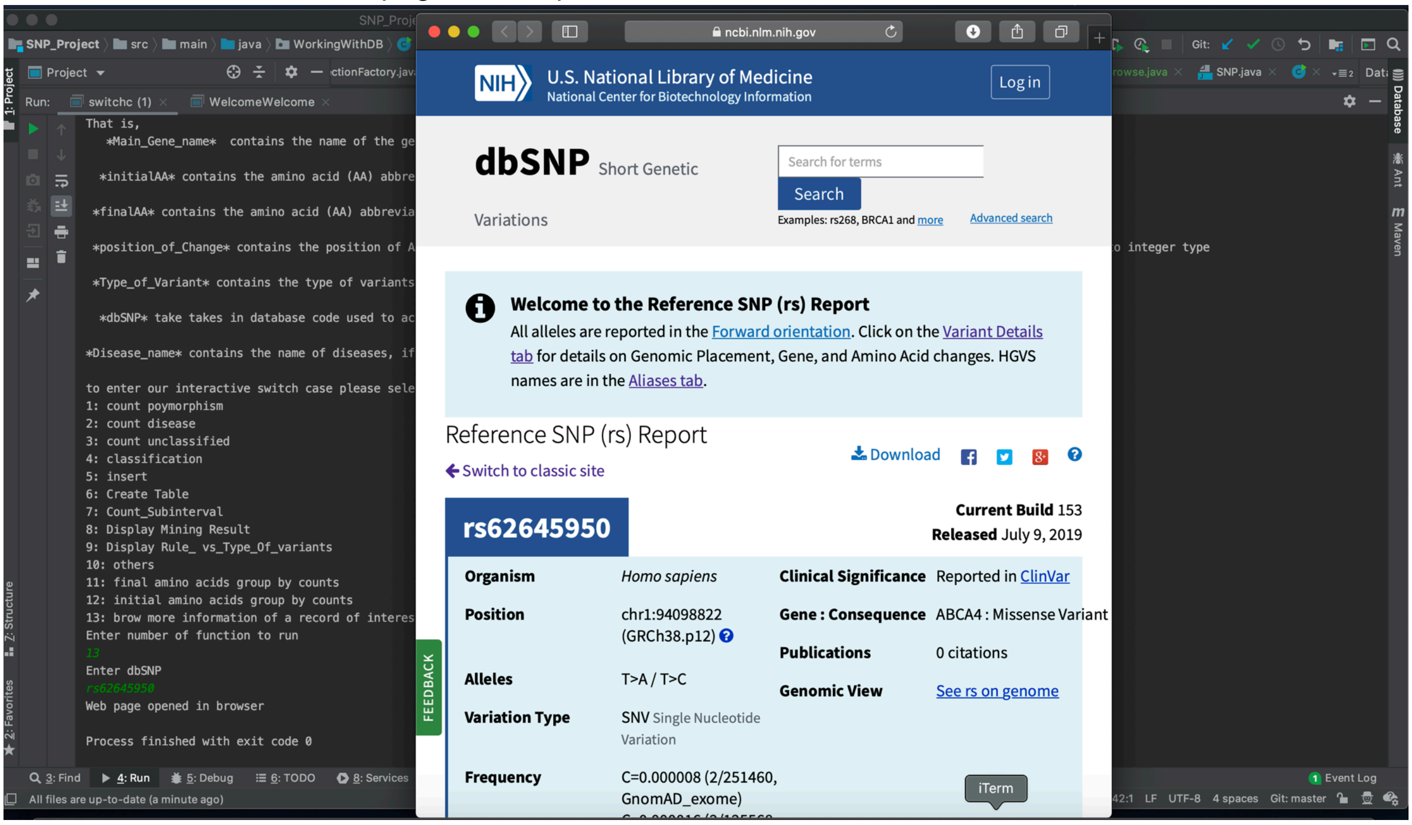
This case counts the initla amino acids: and again, this case should have been the 11th case, i guess....



```
13: brow more information of a record of interest
Enter number of function to run
12
Connected to database
initialaa: Ala count: 5926
initialaa: Arg count: 11612
initialaa: Asn count: 2653
initialaa: Asp count: 3616
initialaa: Cys count: 2598
initialaa: Gln count: 2264
initialaa: Glu count: 3921
initialaa: Gly count: 6720
initialaa: GRA count: 1
initialaa: his count: 1
initialaa: His count: 2156
initialaa: Ile count: 3297
initialaa: Leu count: 4918
initialaa: Lys count: 2324
initialaa: Met count: 2153
initialaa: Phe count: 1763
initialaa: Pro count: 4799
initialaa: Sec count: 1
initialaa: Ser count: 5160
initialaa: Thr count: 4368
initialaa: Trp count: 993
initialaa: Tyr count: 1935
initialaa: Val count: 5067

Process finished with exit code 0
```

Case 13: brow more information of a record of interest

This case allows rthe user to browse more information regarding the record of interest opening the brower with NCBI database page of the specified record.



```
That is,
   *Main_Gene_name*  contains the name of the ge
  *initialAA* contains the amino acid (AA) abbre
 *finalAA* contains the amino acid (AA) abbrevia
 *position_of_Change* contains the position of A
 *Type_of_Variant* contains the type of variants
  *dbSNP* take takes in database code used to ac
*Disease_name* contains the name of diseases, if

to enter our interactive switch case please sele
1: count poymorphism
2: count disease
3: count unclassified
4: classification
5: insert
6: Create Table
7: Count_Subinterval
8: Display Mining Result
9: Display Rule_ vs_Type_Of_variants
10: others
11: final amino acids group by counts
12: initial amino acids group by counts
13: brow more information of a record of interes
Enter number of function to run
13
Enter dbSNP
rs62645950
Web page opened in browser

Process finished with exit code 0
```

U.S. National Library of Medicine
National Center for Biotechnology Information

dbSNP Short Genetic Variations

Examples: rs268, BRCA1 and more Advanced search

Welcome to the Reference SNP (rs) Report
All alleles are reported in the Forward orientation. Click on the Variant Details tab for details on Genomic Placement, Gene, and Amino Acid changes. HGVS names are in the Aliases tab.

Reference SNP (rs) Report

rs62645950

Current Build 153
Released July 9, 2019

Organism	*Homo sapiens*	**Clinical Significance**	Reported in ClinVar
Position	chr1:94098822 (GRCh38.p12)	**Gene : Consequence**	ABCA4 : Missense Variant
		Publications	0 citations
Alleles	T>A / T>C	**Genomic View**	See rs on genome
Variation Type	SNV Single Nucleotide Variation		
Frequency	C=0.000008 (2/251460, GnomAD_exome)		

End of Demonstration

Thank you for you attention, please stay safe!

```
In [ ]:
```