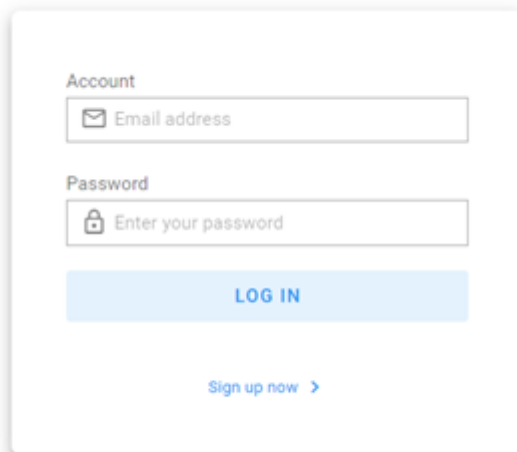


輔助臨床診斷使用之軟體系統，組成架構應達成以下幾項功能：

- NoSQL 資料庫系統，替需永久儲存的分析資料進行存放與管理，
- 運算核心
- 診斷與分析時的圖形化使用者界面，包含了使用者登入、分析樣本建立與上傳、分析進度呈現、分析結果檢索與呈現等功能。
- 容器化建置與佈署：透過 Docker 之容器化技術，上述模組將分別進行所需之編譯、安裝、以及啟用與常駐。

圖形化動態使用者界面

使用者管理



Account

Email address

Password

Enter your password

LOG IN

[Sign up now >](#)

資料皆源自於各檢體之分析資料上傳。

LOGOUT test

SAMPLE

ADD SAMPLE

10 page 1 5 6 record

| Name | Phenotype | Sex | Clinical info | Onset | Severe | Progress | Action |
|----------|--|-----|---------------|-------|--------|----------|--------|
| gerger | Jaundice,Dolichocephaly,Abnormality of the urinary system, Renal insufficiency | ♀ F | rggreg | | | ✓✓✓✓✓ | |
| tsst | Renal insufficiency, Dolichocephaly | ♀ F | | | | ✓✓✓✓✓ | |
| tssttsst | Renal insufficiency, Dolichocephaly | ♀ F | | | | ✓✓✓✓✓ | |
| tssttsst | Renal insufficiency, Dolichocephaly | ♀ F | | | | ✓✓✓✓✓ | |
| tssttsst | Renal insufficiency, Dolichocephaly | ♀ F | | | | ✓✓✓✓✓ | |
| tssttsst | Renal insufficiency, Abnormality of the eye | ♀ F | | | | ✓✓✓✓✓ | |

新增分析檢體資料

Add Sample

File input

Name

Sex

☐ Male ☐ Female

Phenotype

分析進度能透過圖形化介面進行顯示

| Name | Phenotype | Sex | Clinical info | Onset | Severe | Progress | Action |
|--------|--|-----|---------------|-------|--------|----------|--------|
| gerger | Jaundice,Dolichocephaly,Abnormality of the urinary system, Renal insufficiency | ♀ F | rggreg | | | ✓✓✓✓✓ | |

分析結果

RESULT

NORMAL VIEW

PRO VIEW

Result

Phenotype

Panel

MIR3926-1

Variant Amount: 3

MAMDC2-AS1

Variant Amount: 1

CYRIB

Variant Amount: 1

HOXC10

Variant Amount: 1

MIR3926-1

Chr8: 12,727,232 - 12,727,304

Links

Chromosome

Start

End

Assembly

Strand

MIR3926-1 Description

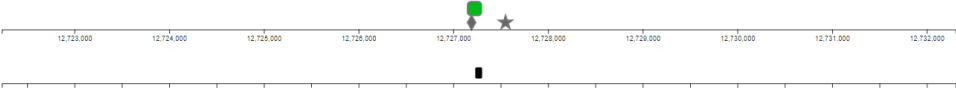
microRNAs (miRNAs) are short (20-24 nt) non-coding RNAs that are involved in post-transcriptional regulation of gene expression in multicellular organisms by affecting both the stability and translation of mRNAs. miRNAs are transcribed by RNA polymerase II as part of capped and polyadenylated primary transcripts (pri-miRNAs) that can be either protein-coding or non-coding. The primary transcript is cleaved by the Drosha ribonuclease III enzyme to produce an approximately 70-nt stem-loop precursor miRNA (pre-miRNA), which is further cleaved by the cytoplasmic Dicer ribonuclease to generate the mature miRNA and antisense miRNA star (miRNA*) products. The mature miRNA is incorporated into a RNA-induced silencing complex (RISC), which recognizes target mRNAs through imperfect base pairing with the miRNA and most commonly results in translational inhibition or destabilization of the target mRNA. The RefSeq represents the predicted microRNA stem-loop. [provided by RefSeq, Sep 2009]

MIR3926-1-201 Variant

Transcript

ENST00000578598.1

MIR3926-1-201 Variant



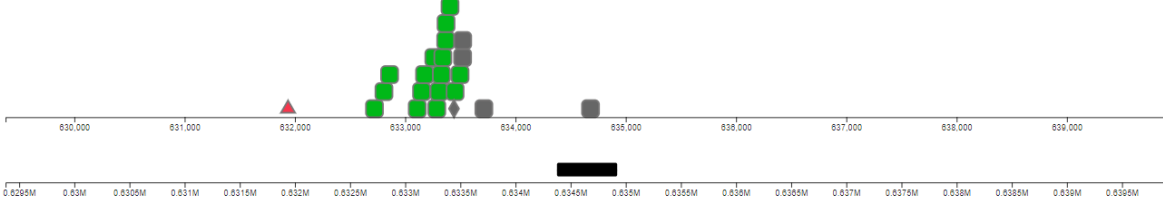
對於選定之 Transcript，得以展示其經過 Sherlock 分析之圖形與相關資訊：

MTCO3P12-201 Variant

Transcript

ENST00000416718.2

MTCO3P12-201 Variant



Sherloc

Tree

MTCO3P12 Variant

HGVSc: — HGVSp: — HGVSG: chr1:g.631862del

REPORT

Pathogenicity

Likely Pathogenic

Population Frequency

GnomAD Genomes

AF < 0.0005

Inheritance Pattern

AD:DVD

Vcf Format

GT: 0/1

AD: 30,24

DP: 54

GQ: 99

PL: 668,0,771

Prediction Tools

CADD PHRED: 26.0

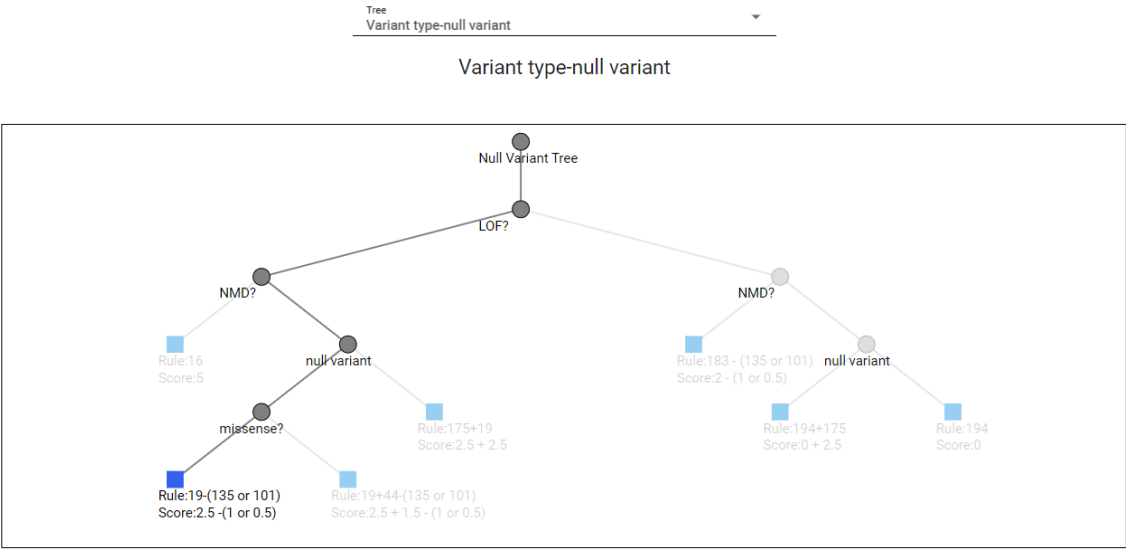
PolyPhen: possibly_damaging(0.856)

REVEL: 0.850

SIFT: deleterious(0.02)

MaxEntScan diff:

pLI gene value: 0.00



使用者彙整自己的基因列表

PANEL

Q

ADD PANEL

| # | Name | Genes | Action |
|---|----------|---------------------------------------|---|
| 1 | ddddsss | RG54,RLN1,RG56,PDE4DIPB,EIF1AXP1,RG55 | MODIFY DELETE |
| 2 | ddd | RG54 | MODIFY DELETE |
| 3 | www | ASIC1,NMNAT2,NMNAT1,DNMT1 | MODIFY DELETE |
| 4 | col pink | COL3A1,SPINK5,MTCO3P12 | MODIFY DELETE |