

# Package ‘EDIRquery’

January 30, 2025

**Title** Query the EDIR Database For Specific Gene

**Version** 1.6.0

## Description

EDIRquery provides a tool to search for genes of interest within the Exome Database of Interspersed Repeats (EDIR). A gene name is a required input, and users can additionally specify repeat sequence lengths, minimum and maximum distance between sequences, and whether to allow a 1-bp mismatch. Outputs include a summary of results by repeat length, as well as a dataframe of query results. Example data provided includes a subset of the data for the gene GAA (ENSG00000171298). To query the full database requires providing a path to the downloaded database files as a parameter.

**License** GPL-3

**Encoding** UTF-8

**biocViews** Genetics, SequenceMatching

**Roxygen** list(markdown = TRUE)

**RoxygenNote** 7.2.3

**Depends** R (>= 4.2.0)

**Imports** tibble (>= 3.1.6), tictoc (>= 1.0.1), utils (>= 4.1.3), stats (>= 4.1.3), readr (>= 2.1.2), InteractionSet (>= 1.22.0), GenomicRanges (>= 1.46.1)

**Suggests** knitr, rmarkdown, testthat (>= 3.0.0)

**VignetteBuilder** knitr

**Config/testthat/edition** 3

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## Contents

|                       |   |
|-----------------------|---|
| gene_chr . . . . .    | 2 |
| gene_lookup . . . . . | 2 |

**Index****4**

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|          |                                 |
|----------|---------------------------------|
| gene_chr | <i>Gene chromosome location</i> |
|----------|---------------------------------|

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**Description**

A dataset containing two formats of gene names and associated chromosome number.

**Usage**

```
gene_chr
```

**Format**

A data frame with 60571 rows and 3 variables:

**chromosome\_name** chromosome

**ensembl\_gene\_id** Ensembl gene ID of gene

**hgnc\_symbol** HGNC symbol of gene

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|             |                                       |
|-------------|---------------------------------------|
| gene_lookup | <i>Look Up a Gene in EDIR Dataset</i> |
|-------------|---------------------------------------|

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**Description**

This function searches for a specified gene in the EDIR dataset. A gene name is a required parameter.

**Usage**

```
gene_lookup(  
  gene,  
  length = NA,  
  mindist = 0,  
  maxdist = 1000,  
  format = "data.frame",  
  summary = FALSE,  
  mismatch = TRUE,  
  path = NA  
)
```

## Arguments

|          |   |
|----------|---|
| gene     | The gene name (ENSEMBL ID or HGNC symbol)   |
| length   | Repeat sequence length, must be between 7 and 20. Defaults to NA. If NA, results will include all available lengths in dataset for queried gene.                              |
| mindist  | Minimum spacer distance between repeats. Defaults to 0.   |
| maxdist  | Maximum spacer distance between repeats. Defaults to 1000.  |
| format   | Output table format. One of 'data.frame', 'GInteractions'. Defaults to 'data.frame'.  |
| summary  | Logical value indicating whether to store summary. Defaults to FALSE.   |
| mismatch | Logical value indicating whether to allow 1 mismatch in sequences. Defaults to TRUE.  |
| path     | String containing path to directory holding downloaded dataset files. Defaults to NA. If not provided (path = NA), gene_lookup() will use subset of data provided as example. |

## Details

Summary of results printed to console includes gene name, gene length (bp), Ensembl transcript ID, queried distance between repeats (default: 0-1000 bp), and an overview of total results for the given repeat length. Console outputs include runtime.

## Value

A data.frame of the results from the EDIR database. If summary = TRUE, returns a tibble containing the summary (\$summary), and query results (\$results).

## Examples

```
## With given repeat length,
gene_lookup("GAA", length = 7, mindist = 10, maxdist = 1000,
            mismatch = TRUE)

## Without specified repeat length
gene_lookup("GAA", mindist = 0, maxdist = 1000, mismatch = TRUE)

## To access query results, store in variable
output <- gene_lookup("GAA", length = 7, mindist = 10, maxdist = 1000,
                    mismatch = FALSE)
head(output)

## With summary = TRUE
output <- gene_lookup("GAA", length = 10, mindist = 10, maxdist = 1000,
                    summary = TRUE,
                    mismatch = TRUE)

output$summary
head(output$results)
```

# Index

## \* datasets

gene\_chr, [2](#)

gene\_chr, [2](#)

gene\_lookup, [2](#)