

TARGET-seq genotyped single-cell RNA sequencing of hematopoietic stem cells and megakaryocyte-erythroid progenitor cells from a dual SF3B1-mutant MDS-RS patient and 3 healthy donors

SND-ID: 2023-223. **Version:** 1. **DOI:** <https://doi.org/10.48723/d7s9-6336>

Associated documentation

DualMT_TARGETseq_FileList.txt (2.25 KB)

Readme_TARGETseq.txt (1.24 KB)

Citation

Moura, P. L., & Hellström-Lindberg, E. (2024) TARGET-seq genotyped single-cell RNA sequencing of hematopoietic stem cells and megakaryocyte-erythroid progenitor cells from a dual SF3B1-mutant MDS-RS patient and 3 healthy donors (Version 1) [Data set]. Karolinska Institutet. Available at: <https://doi.org/10.48723/d7s9-6336>

Creator/Principal investigator(s)

[Pedro Luis Moura](#) - Karolinska Institutet, Department of Medicine, Huddinge / Center for Hematology and Regenerative Medicine (HERM)

[Eva Hellström-Lindberg](#) - Karolinska Institutet, Department of Medicine, Huddinge / Center for Hematology and Regenerative Medicine (HERM)

Research principal

[Karolinska Institutet](#) - Department of Medicine, Huddinge / Center for Hematology and Regenerative Medicine (HERM)

Description

This dataset consists of TARGETseq (genotype-targeted plate-based SmartSeq2) single-cell RNA sequencing data of purified hematopoietic stem cells (HSC) and megakaryocyte-erythroid progenitors (MEP) from the bone marrow of a dual SF3B1-mutant MDS-RS patient (Patient 1 in the associated publication) over 2 timepoints (39 months post-diagnosis, with dominant SF3B1-N626D, and 118 months post-diagnosis, with dominant SF3B1-K666N); and purified HSC/MEP from the bone marrow of three healthy donors. The objective of this data collection was to assess the molecular characteristics that increase fitness in SF3B1-mutant HSC as compared to normal HSC.

The dataset is approximately 187 GB and includes the file types: zip, xlsx, bam, bam.bai, rds.

Data contains personal data

Yes

Sensitive personal data

Yes

Type of personal data

Genetic and biological data of patients

Code key exists

Yes

Language

[English](#)

Unit of analysis

[Cells](#)

Population

Patients with Myelodysplastic neoplasms with ring sideroblasts (MDS-RS)

Time Method

[Time series: Discrete](#)

Study design

Preclinical study

Sampling procedure

[Probability: Simple random](#)

[Non-probability: Availability](#)

[Mixed probability and non-probability](#)

The data in this dataset comes from 1 dual SF3B1mt MDS-RS patient and 3 healthy donors. Bone marrow (BM) samples were collected from 2 dual SF3B1mt MDS-RS patients (Patient 1: N626D, K666N; Patient 2: K700E, K666N) evaluated at Karolinska University Hospital, Sweden. Diagnostic procedures were performed according to the European LeukemiaNet recommendation and WHO classification for myeloid neoplasms. Mutational status was evaluated at the clinic through panel sequencing for the most common myeloid mutations. Additional samples were collected from a total of 4 healthy NBM donors for control purposes. All source material was provided with written informed consent for research use, given in accordance with the Declaration of Helsinki, and the study was approved by the Ethics Research Committee at Karolinska Institutet.

Biobank is connected to the study

This study has used existing samples from a scientific collection or biobank

Scientific collection or biobank name: Karolinska Institutet MDS biobank

Type(s) of sample: Bone marrow cells

Data format / data structure

[Numeric](#)

[Other](#)

Responsible department/unit

Department of Medicine, Huddinge / Center for Hematology and Regenerative Medicine (HERM)

Contributor(s)

Affaf Aliouat - University of Oxford

Sten Eirik Jacobsen - Karolinska Institutet

Petter Woll - Karolinska Institutet

Funding 1

- Funding agency: Swedish Cancer Society
- Funding agency's reference number: 21 0340

Funding 2

- Funding agency: Swedish Research Council
- Funding agency's reference number: 211133

Funding 3

- Funding agency: Knut and Alice Wallenberg Foundation
- Funding agency's reference number: 2017.0359

Funding 4

- Funding agency: Swedish Cancer Society
- Funding agency's reference number: 19 0200

Ethics Review

Stockholm - Ref. 2017/1090-31/4

Research area

[Natural sciences](#) (Standard för svensk indelning av forskningsämnen 2011)

[Biological sciences](#) (Standard för svensk indelning av forskningsämnen 2011)

[Cell biology](#) (Standard för svensk indelning av forskningsämnen 2011)

[Genetics](#) (Standard för svensk indelning av forskningsämnen 2011)

[Bioinformatics and systems biology](#) (Standard för svensk indelning av forskningsämnen 2011)

[Hematology](#) (Standard för svensk indelning av forskningsämnen 2011)

Keywords

[Mds-rs](#), [Sf3b1](#), [Myelodysplastic syndromes](#)

Publications

Pedro Luis Moura, Yasuhito Nannya, Affaf Aliouat, Isabel Juliana Hofman, Teresa Mortera Blanco, Tetsuichi Yoshizato, Ryunosuke Saiki, Masahiro M Nakagawa, Maria Creignou, Ann-Charlotte Björklund, Gunilla Walldin, Indira Barbosa, Monika Jansson, Francesca Grasso, Edda M Elvarsdottir, Petter S Woll, Sten Eirik W Jacobsen, Seishi Ogawa, Eva Hellström-Lindberg, Competition of dual SF3B1mt clones in MDS-RS is associated with distinct RNA mis-splicing in hematopoietic stem cells, Blood Neoplasia, 2024,100011, ISSN 2950-3280,

<https://doi.org/10.1016/j.bneo.2024.100011>.

DOI: <https://doi.org/10.1016/j.bneo.2024.100011>

Accessibility level

Access to data through SND

Access to data is restricted

Use of data

[Things to consider when using data shared through SND](#)

License

[CC BY-NC-ND 4.0](#)

Versions

Version 1. 2024-04-18

Download metadata

[DataCite](#)

[DDI 2.5](#)

[DDI 3.3](#)

[DCAT-AP-SE 2.0](#)

[JSON-LD](#)

[PDF](#)

[Citation \(CSL\)](#)

Published: 2024-04-18