

## Editorial Policy Checklist

This form is used to ensure compliance with Nature Portfolio editorial policies related to research ethics and reproducibility. For further information, please see our [editorial policies](#) site. All relevant questions on the form must be answered.

### Competing interests

Policy information about [competing interests](#)

#### Competing interests declaration

In the interest of transparency and to help readers form their own judgements of potential bias, Nature Portfolio journals require authors to declare any competing financial and/or non-financial interest in relation to the work described in the submitted manuscript.

- ☐ We declare that none of the authors have competing financial or non-financial interests as defined by Nature Portfolio.
- ☒ We declare that one or more of the authors have a competing interest as defined by Nature Portfolio.

Dr. Green has received compensation for advising Genomic Life and is co-founder of Genome Medical.  
 Dr. Shah is a member of the Scientific Advisory Board for Neuberger Center for Genomic Medicine.  
 Ms. Genetti has received compensation for consulting for Kate Therapeutics.  
 Dr. Yu has consulted and received compensation or honoraria from Eisai, BioMarin, GeneTx, Takeda, and Alnylam.  
 Ms. Zettler has received compensation for consulting for Novartis Gene Therapies.  
 Dr. Lebo is employed by a not-for-profit, fee-for-service clinical laboratory at Mass General Brigham offering genomic screening.  
 Dr. Agrawal is a member of the Scientific Advisory Board for GeneDx and Illumina, Inc.  
 Dr. McGuire is a member of the board of the Greenwall Foundation and is on the Scientific Advisory Board for Danaher Life Sciences, Geisinger Research, and the Morgridge Institute for Research.  
 Dr. Rehm is a member of the Scientific Advisory Board for Genome Medical and is employed as the medical and clinical laboratory director for a fee-for service laboratory offering genomic sequencing at the Broad Institute of MIT and Harvard.  
 Dr. Holm is a member of the Scientific Advisory Board for Biomarin for vorosoritide.  
 Dr. Beggs has received funding from the NIH, MDA (USA), AFM Telethon, Alexion Pharmaceuticals Inc., Audentes Therapeutics Inc., Avidity Biosciences, Dynacure SAS, and Pfizer Inc. He has consulted and received compensation or honoraria from Asklepios BioPharmaceutical Inc, Audentes Therapeutics, Biogen, F. Hoffman-La Roche AG, GLG Inc, Guidepoint Global, and Kate Therapeutics, and holds equity in Kinea Biosciences and Kate Therapeutics.

### Authorship

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Prior to submission all listed authors must agree to all manuscript contents, the author list and its order and the author contribution statements. Any changes to the author list after submission must be approved by all authors.

- ☒ We have read the Nature Portfolio Authorship Policy and confirm that this manuscript complies.

### Data availability

Policy information about [availability of data](#)

#### Data availability statement

All manuscripts must include a [data availability statement](#). This statement should provide the following information, where applicable:

- Accession codes, unique identifiers, or web links for publicly available datasets
- A description of any restrictions on data availability
- For clinical datasets or third party data, please ensure that the statement adheres to our [policy](#)

- ☒ We have provided a full data availability statement in the manuscript.

**Mandated accession codes** ([where applicable](#))

Confirm that all relevant data are deposited into a public repository and that accession codes are provided.

☒ All relevant accession codes are provided ☐ Accession codes will be available before publication ☐ No data with mandated deposition

**Code availability**

Policy information about [availability of computer code](#)

**Code availability statement**

For all studies using custom code or mathematical algorithm that is deemed central to the conclusions, the manuscript must include a statement under the heading "Code availability" describing how readers can access the code, including any access restrictions. Code availability statements should be provided as a separate section after the data availability statement but before the References.

☒ We have provided a full code availability statement in the manuscript

**Data presentation**

For all data presented in a plot, chart or other visual representation confirm that:

n/a | Confirmed

- ☐ ☒ Individual data points are shown when possible, and always for  $n \leq 10$
- ☒ ☐ The format shows data distribution clearly (e.g. dot plots, box-and-whisker plots)
- ☒ ☐ Box-plot elements are defined (e.g. center line, median; box limits, upper and lower quartiles; whiskers, 1.5x interquartile range; points, outliers)
- ☒ ☐ Clearly defined error bars are present and what they represent (SD, SE, CI) is noted

**Image integrity**

Policy information about [image integrity](#)

☒ We have read Nature Portfolio's image integrity policy and all images comply.

Unprocessed data must be provided upon request. Please double-check figure assembly to ensure that all panels are accurate (e.g. all labels are correct, no inadvertent duplications have occurred during preparation, etc.).

Where blots and gels are presented, please take particular care to ensure that lanes have not been spliced together, that loading controls are run on the same blot, and that unprocessed scans match the corresponding figures.

**Additional policy considerations**

Some types of research require additional policy disclosures. Please indicate whether each of these apply to your study. If you are not certain, please read the appropriate section before selecting a response.

Does not apply	Involved in the study
<input checked="" type="checkbox"/>	<input type="checkbox"/> Macromolecular structural data
<input checked="" type="checkbox"/>	<input type="checkbox"/> Unique biological materials
<input checked="" type="checkbox"/>	<input type="checkbox"/> Research animals and/or animal-derived materials that require ethical approval
<input checked="" type="checkbox"/>	<input type="checkbox"/> Human embryos, gametes and/or stem cells
<input type="checkbox"/>	<input checked="" type="checkbox"/> Human research participants
<input type="checkbox"/>	<input checked="" type="checkbox"/> Clinical data
<input checked="" type="checkbox"/>	<input type="checkbox"/> Archaeological, geological, and palaeontological materials

## Human research participants

Policy information about [studies involving human research participants](#)

### Ethical compliance

☒ We have complied with all relevant ethical regulations and include a statement affirming this in the manuscript.

### Ethics committee

Confirm that the manuscript states the name(s) of the board and/or institution that:

☒ Approved the study protocol -OR- ☐ Provided guidelines for study procedures (if protocol approval is not required)

### Informed consent

☒ We have obtained informed consent from all participants and this is noted in the manuscript.

### Identifiable images

For publication of identifiable images of research participants, confirm that consent to publish was obtained and is noted in the Methods.

Authors must ensure that consent meets the conditions set out in the [Nature Portfolio participant release form](#).

☐ Yes ☒ No identifiable images of human research participants

## Clinical studies

Policy information about [clinical studies](#)

### Clinical trial registration

☒ We have provided the trial registration number from [ClinicalTrials.gov](#) or an equivalent agency in the manuscript.

### Phase 2 and 3 randomized controlled trials

We have provided the [CONSORT checklist](#) with your submission.

☐ Yes ☐ No ☒ Not a phase 2/3 randomized controlled trial

### Tumor marker prognostic studies

We have followed the [REMARK reporting guidelines](#).

☐ Yes ☐ No ☒ Not a tumor marker prognostic study

I certify that all the above information is complete and correct.

Typed signature Robert C Green

Date Mar 17, 2022