

ICMJE Form for Disclosure of Potential Conflicts of Interest

Instructions

The purpose of this form is to provide readers of your manuscript with information about your other interests that could influence how they receive and understand your work. The form is designed to be completed electronically and stored electronically. It contains programming that allows appropriate data display. Each author should submit a separate form and is responsible for the accuracy and completeness of the submitted information. The form is in six parts.

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This section asks for information about the work that you have submitted for publication. The time frame for this reporting is that of the work itself, from the initial conception and planning to the present. The requested information is about resources that you received, either directly or indirectly (via your institution), to enable you to complete the work. Checking "No" means that you did the work without receiving any financial support from any third party -- that is, the work was supported by funds from the same institution that pays your salary and that institution did not receive third-party funds with which to pay you. If you or your institution received funds from a third party to support the work, such as a government granting agency, charitable foundation or commercial sponsor, check "Yes".

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4. Intellectual Property.

This section asks about patents and copyrights, whether pending, issued, licensed and/or receiving royalties.

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Other: Anything not covered under the previous three boxes

Pending: The patent has been filed but not issued

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Licensed: The patent has been licensed to an entity, whether earning royalties or not

Royalties: Funds are coming in to you or your institution due to your patent

ICMJE Form for Disclosure of Potential Conflicts of Interest

Section 1. Identifying Information

| | | |
|---|----------------------------------|----------------------------|
| 1. Given Name (First Name) James | 2. Surname (Last Name) Lupski | 3. Date 26-October-2015 |
| 4. Are you the corresponding author? <input checked="" type="checkbox"/> Yes <input type="checkbox"/> No | | |
| 5. Manuscript Title Molecular etiology of arthrogyrosis in a cohort of families of mostly Turkish origin | | |
| 6. Manuscript Identifying Number (if you know it) 84457-RG-1 | | |

Section 2. The Work Under Consideration for Publication

Did you or your institution **at any time** receive payment or services from a third party (government, commercial, private foundation, etc.) for any aspect of the submitted work (including but not limited to grants, data monitoring board, study design, manuscript preparation, statistical analysis, etc.)?

Are there any relevant conflicts of interest? ☒ Yes ☐ No

If yes, please fill out the appropriate information below. If you have more than one entity press the "ADD" button to add a row. Excess rows can be removed by pressing the "X" button.

| Name of Institution/Company | Grant? | Personal Fees? | Non-Financial Support? | Other? | Comments |
|-----------------------------|-------------------------------------|--------------------------|--------------------------|--------------------------|--------------|
| NIH/NINDS | <input checked="" type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | U54 HG006542 |

Section 3. Relevant financial activities outside the submitted work.

Place a check in the appropriate boxes in the table to indicate whether you have financial relationships (regardless of amount of compensation) with entities as described in the instructions. Use one line for each entity; add as many lines as you need by clicking the "Add +" box. You should report relationships that were **present during the 36 months prior to publication**.

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| Name of Entity | Grant? | Personal Fees? | Non-Financial Support? | Other? | Comments |
|----------------|--------------------------|--------------------------|--------------------------|-------------------------------------|-----------------|
| 23andMe | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input checked="" type="checkbox"/> | Stock ownership |
| Regeneron | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input checked="" type="checkbox"/> | Consultant |
| Lasergen | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input checked="" type="checkbox"/> | Stock options |

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| Name of Entity | Grant? | Personal Fees? | Non-Financial Support? | Other? | Comments |
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| Miraca Genetics Laboratories | <input type="checkbox"/> | <input type="checkbox"/> | <input checked="" type="checkbox"/> | <input type="checkbox"/> | Scientific Advisory Board |

Section 4. Intellectual Property -- Patents & Copyrights

Do you have any patents, whether planned, pending or issued, broadly relevant to the work? ☐ Yes ☒ No

Section 5. Relationships not covered above

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Dr. Lupski reports grants from NIH/NINDS , during the conduct of the study; other from 23andMe, other from Regeneron, other from Lasergen, non-financial support from Miraca Genetics Laboratories, outside the submitted work; .

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Section 1. Identifying Information

| | | |
|---|---------------------------------|--|
| 1. Given Name (First Name) Richard | 2. Surname (Last Name) Gibbs | 3. Date 30-October-2015 |
| 4. Are you the corresponding author? <input type="checkbox"/> Yes <input checked="" type="checkbox"/> No | | Corresponding Author's Name James R. Lupski |
| 5. Manuscript Title Molecular etiology of arthrogyposis in a cohort of families of mostly Turkish origin | | |
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| Miraca Genetics Laboratories | <input type="checkbox"/> | <input type="checkbox"/> | <input checked="" type="checkbox"/> | <input type="checkbox"/> | Interim Chief Scientific Officer |

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Section 1. Identifying Information

1. Given Name (First Name)
Yavuz

2. Surname (Last Name)
Bayram

3. Date
30-October-2015

4. Are you the corresponding author?

☐ Yes ☒ No

Corresponding Author's Name
James R. Lupski

5. Manuscript Title
Molecular etiology of arthrogryposis in a cohort of families of mostly Turkish origin

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Dr. Bayram has nothing to disclose.

Evaluation and Feedback

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Section 1. Identifying Information

| | | |
|--|----------------------------------|--|
| 1. Given Name (First Name) Ender | 2. Surname (Last Name) Karaca | 3. Date 30-October-2015 |
| 4. Are you the corresponding author? <input type="checkbox"/> Yes <input checked="" type="checkbox"/> No | | Corresponding Author's Name James R. Lupski |
| 5. Manuscript Title Molecular etiology of arthrogryposis in a cohort of families of mostly Turkish origin | | |
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1. Given Name (First Name)
Zeynep

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Coban Akdemir

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☐ Yes ☒ No

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ICMJE Form for Disclosure of Potential Conflicts of Interest

Section 1. Identifying Information

1. Given Name (First Name)
Elif

2. Surname (Last Name)
Ozdamar Yilmaz

3. Date
30-October-2015

4. Are you the corresponding author?

☐ Yes ☒ No

Corresponding Author's Name
James R. Lupski

5. Manuscript Title
Molecular etiology of arthrogryposis in a cohort of families of mostly Turkish origin

6. Manuscript Identifying Number (if you know it)
84457-RG-1

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Dr. Ozdamar Yilmaz has nothing to disclose.

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ICMJE Form for Disclosure of Potential Conflicts of Interest

Section 1. Identifying Information

1. Given Name (First Name)
Gulsen

2. Surname (Last Name)
Akay Tayfun

3. Date
30-October-2015

4. Are you the corresponding author?

☐ Yes ☒ No

Corresponding Author's Name
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5. Manuscript Title
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Dr. Akay Tayfun has nothing to disclose.

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Section 1. Identifying Information

1. Given Name (First Name)
Hatip

2. Surname (Last Name)
Aydin

3. Date
30-October-2015

4. Are you the corresponding author?

☐ Yes

☒ No

Corresponding Author's Name
James R. Lupski

5. Manuscript Title
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Dr. Aydin has nothing to disclose.

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Section 1. Identifying Information

| | | |
|--|---------------------------------|--|
| 1. Given Name (First Name) Deniz | 2. Surname (Last Name) Torun | 3. Date 30-October-2015 |
| 4. Are you the corresponding author? <input type="checkbox"/> Yes <input checked="" type="checkbox"/> No | | Corresponding Author's Name James R. Lupski |
| 5. Manuscript Title Molecular etiology of arthrogryposis in a cohort of families of mostly Turkish origin | | |
| 6. Manuscript Identifying Number (if you know it) 84457-RG-1 | | |

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Dr. Torun has nothing to disclose.

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Section 1. Identifying Information

1. Given Name (First Name)
Sevcan

2. Surname (Last Name)
Tug Bozdogan

3. Date
30-October-2015

4. Are you the corresponding author?

☐ Yes

☒ No

Corresponding Author's Name
James R. Lupski

5. Manuscript Title
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Dr. Tug Bozdogan has nothing to disclose.

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Royalties: Funds are coming in to you or your institution due to your patent

ICMJE Form for Disclosure of Potential Conflicts of Interest

Section 1. Identifying Information

| | | |
|--|-------------------------------------|--|
| 1. Given Name (First Name) Alper | 2. Surname (Last Name) Gezdirici | 3. Date 30-October-2015 |
| 4. Are you the corresponding author? <input type="checkbox"/> Yes <input checked="" type="checkbox"/> No | | Corresponding Author's Name James R. Lupski |
| 5. Manuscript Title Molecular etiology of arthrogryposis in a cohort of families of mostly Turkish origin | | |
| 6. Manuscript Identifying Number (if you know it) 84457-RG-1 | | |

Section 2. The Work Under Consideration for Publication

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Are there any relevant conflicts of interest? ☐ Yes ☒ No

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Are there any relevant conflicts of interest? ☐ Yes ☒ No

Section 4. Intellectual Property -- Patents & Copyrights

Do you have any patents, whether planned, pending or issued, broadly relevant to the work? ☐ Yes ☒ No

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Section 6. Disclosure Statement

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Dr. Gezdirici has nothing to disclose.

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ICMJE Form for Disclosure of Potential Conflicts of Interest

Section 1. Identifying Information

| | | |
|--|----------------------------------|--|
| 1. Given Name (First Name) Sedat | 2. Surname (Last Name) Isikay | 3. Date 30-October-2015 |
| 4. Are you the corresponding author? <input type="checkbox"/> Yes <input checked="" type="checkbox"/> No | | Corresponding Author's Name James R. Lupski |
| 5. Manuscript Title Molecular etiology of arthrogryposis in a cohort of families of mostly Turkish origin | | |
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Are there any relevant conflicts of interest? ☐ Yes ☒ No

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Dr. Isikay has nothing to disclose.

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ICMJE Form for Disclosure of Potential Conflicts of Interest

Section 1. Identifying Information

1. Given Name (First Name)
Mehmed

2. Surname (Last Name)
Atik

3. Date
30-October-2015

4. Are you the corresponding author?

☐ Yes ☒ No

Corresponding Author's Name
James R. Lupski

5. Manuscript Title
Molecular etiology of arthrogryposis in a cohort of families of mostly Turkish origin

6. Manuscript Identifying Number (if you know it)
84457-RG-1

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ICMJE Form for Disclosure of Potential Conflicts of Interest

Section 1. Identifying Information

1. Given Name (First Name)

Tomasz

2. Surname (Last Name)

Gambin

3. Date

30-October-2015

4. Are you the corresponding author?

☐ Yes

☒ No

Corresponding Author's Name

James R. Lupski

5. Manuscript Title

Molecular etiology of arthrogryposis in a cohort of families of mostly Turkish origin

6. Manuscript Identifying Number (if you know it)

84457-RG-1

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ICMJE Form for Disclosure of Potential Conflicts of Interest

Section 1. Identifying Information

| | | |
|--|---------------------------------|--|
| 1. Given Name (First Name) Tamar | 2. Surname (Last Name) Harel | 3. Date 30-October-2015 |
| 4. Are you the corresponding author? <input type="checkbox"/> Yes <input checked="" type="checkbox"/> No | | Corresponding Author's Name James R. Lupski |
| 5. Manuscript Title Molecular etiology of arthrogryposis in a cohort of families of mostly Turkish origin | | |
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Dr. Harel has nothing to disclose.

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ICMJE Form for Disclosure of Potential Conflicts of Interest

Section 1. Identifying Information

| | | |
|--|-------------------------------------|--|
| 1. Given Name (First Name) Ayman | 2. Surname (Last Name) El-Hattab | 3. Date 30-October-2015 |
| 4. Are you the corresponding author? <input type="checkbox"/> Yes <input checked="" type="checkbox"/> No | | Corresponding Author's Name James R. Lupski |
| 5. Manuscript Title Molecular etiology of arthrogryposis in a cohort of families of mostly Turkish origin | | |
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Section 6. Disclosure Statement

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Dr. El-Hattab has nothing to disclose.

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ICMJE Form for Disclosure of Potential Conflicts of Interest

Section 1. Identifying Information

1. Given Name (First Name)
Wu-Lin

2. Surname (Last Name)
Charng

3. Date
30-October-2015

4. Are you the corresponding author?

☐ Yes ☒ No

Corresponding Author's Name
James R. Lupski

5. Manuscript Title
Molecular etiology of arthrogryposis in a cohort of families of mostly Turkish origin

6. Manuscript Identifying Number (if you know it)
84457-RG-1

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Section 1. Identifying Information

1. Given Name (First Name)
Davut

2. Surname (Last Name)
Pehlivan

3. Date
30-October-2015

4. Are you the corresponding author?

☐ Yes

☒ No

Corresponding Author's Name
James R. Lupski

5. Manuscript Title
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| | | |
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| 1. Given Name (First Name) Shalini | 2. Surname (Last Name) Jhangiani | 3. Date 30-October-2015 |
| 4. Are you the corresponding author? <input type="checkbox"/> Yes <input checked="" type="checkbox"/> No | | Corresponding Author's Name James R. Lupski |
| 5. Manuscript Title Molecular etiology of arthrogryposis in a cohort of families of mostly Turkish origin | | |
| 6. Manuscript Identifying Number (if you know it) 84457-RG-1 | | |

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ICMJE Form for Disclosure of Potential Conflicts of Interest

Section 1. Identifying Information

1. Given Name (First Name)
Donna

2. Surname (Last Name)
Muzny

3. Date
30-October-2015

4. Are you the corresponding author? ☐ Yes ☒ No Corresponding Author's Name
James R. Lupski

5. Manuscript Title
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6. Manuscript Identifying Number (if you know it)
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Dr. Muzny has nothing to disclose.

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ICMJE Form for Disclosure of Potential Conflicts of Interest

Section 1. Identifying Information

| | | |
|--|-----------------------------------|--|
| 1. Given Name (First Name) Ali | 2. Surname (Last Name) Karaman | 3. Date 30-October-2015 |
| 4. Are you the corresponding author? <input type="checkbox"/> Yes <input checked="" type="checkbox"/> No | | Corresponding Author's Name James R. Lupski |
| 5. Manuscript Title Molecular etiology of arthrogryposis in a cohort of families of mostly Turkish origin | | |
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Royalties: Funds are coming in to you or your institution due to your patent

ICMJE Form for Disclosure of Potential Conflicts of Interest

Section 1. Identifying Information

1. Given Name (First Name)
Tamer

2. Surname (Last Name)
Celik

3. Date
30-October-2015

4. Are you the corresponding author?

☐ Yes ☒ No

Corresponding Author's Name
James R. Lupski

5. Manuscript Title
Molecular etiology of arthrogryposis in a cohort of families of mostly Turkish origin

6. Manuscript Identifying Number (if you know it)
84457-RG-1

Section 2. The Work Under Consideration for Publication

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Are there any relevant conflicts of interest? ☐ Yes ☒ No

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Are there any relevant conflicts of interest? ☐ Yes ☒ No

Section 4. Intellectual Property -- Patents & Copyrights

Do you have any patents, whether planned, pending or issued, broadly relevant to the work? ☐ Yes ☒ No

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Dr. Celik has nothing to disclose.

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ICMJE Form for Disclosure of Potential Conflicts of Interest

Section 1. Identifying Information

| | | |
|--|---|--|
| 1. Given Name (First Name) Ozge | 2. Surname (Last Name) Ozalp Yuregir | 3. Date 30-October-2015 |
| 4. Are you the corresponding author? <input type="checkbox"/> Yes <input checked="" type="checkbox"/> No | | Corresponding Author's Name James R. Lupski |
| 5. Manuscript Title Molecular etiology of arthrogryposis in a cohort of families of mostly Turkish origin | | |
| 6. Manuscript Identifying Number (if you know it) 84457-RG-1 | | |

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Dr. Ozalp Yuregir has nothing to disclose.

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ICMJE Form for Disclosure of Potential Conflicts of Interest

Section 1. Identifying Information

1. Given Name (First Name)
Timur

2. Surname (Last Name)
Yildirim

3. Date
30-October-2015

4. Are you the corresponding author?

☐ Yes ☒ No

Corresponding Author's Name
James R. Lupski

5. Manuscript Title
Molecular etiology of arthrogryposis in a cohort of families of mostly Turkish origin

6. Manuscript Identifying Number (if you know it)
84457-RG-1

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Dr. Yildirim has nothing to disclose.

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ICMJE Form for Disclosure of Potential Conflicts of Interest

Section 1. Identifying Information

1. Given Name (First Name)
Ilhan

2. Surname (Last Name)
Bayhan

3. Date
30-October-2015

4. Are you the corresponding author?

☐ Yes ☒ No

Corresponding Author's Name
James R. Lupski

5. Manuscript Title
Molecular etiology of arthrogryposis in a cohort of families of mostly Turkish origin

6. Manuscript Identifying Number (if you know it)
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Dr. Bayhan has nothing to disclose.

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ICMJE Form for Disclosure of Potential Conflicts of Interest

Section 1. Identifying Information

1. Given Name (First Name)
Eric

2. Surname (Last Name)
Boerwinkle

3. Date
30-October-2015

4. Are you the corresponding author?

☐ Yes ☒ No

Corresponding Author's Name
James R. Lupski

5. Manuscript Title
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Dr. Boerwinkle has nothing to disclose.

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ICMJE Form for Disclosure of Potential Conflicts of Interest

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| | | |
|--|------------------------------------|--|
| 1. Given Name (First Name) Nursel | 2. Surname (Last Name) Elcioglu | 3. Date 30-October-2015 |
| 4. Are you the corresponding author? <input type="checkbox"/> Yes <input checked="" type="checkbox"/> No | | Corresponding Author's Name James R. Lupski |
| 5. Manuscript Title Molecular etiology of arthrogryposis in a cohort of families of mostly Turkish origin | | |
| 6. Manuscript Identifying Number (if you know it) 84457-RG-1 | | |

Section 2. The Work Under Consideration for Publication

Did you or your institution **at any time** receive payment or services from a third party (government, commercial, private foundation, etc.) for any aspect of the submitted work (including but not limited to grants, data monitoring board, study design, manuscript preparation, statistical analysis, etc.)?

Are there any relevant conflicts of interest? ☐ Yes ☒ No

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Place a check in the appropriate boxes in the table to indicate whether you have financial relationships (regardless of amount of compensation) with entities as described in the instructions. Use one line for each entity; add as many lines as you need by clicking the "Add +" box. You should report relationships that were **present during the 36 months prior to publication**.

Are there any relevant conflicts of interest? ☐ Yes ☒ No

Section 4. Intellectual Property -- Patents & Copyrights

Do you have any patents, whether planned, pending or issued, broadly relevant to the work? ☐ Yes ☒ No

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Section 5. Relationships not covered above

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At the time of manuscript acceptance, journals will ask authors to confirm and, if necessary, update their disclosure statements. On occasion, journals may ask authors to disclose further information about reported relationships.

Section 6. Disclosure Statement

Based on the above disclosures, this form will automatically generate a disclosure statement, which will appear in the box below.

Dr. Elcioglu has nothing to disclose.

Evaluation and Feedback

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Instructions

The purpose of this form is to provide readers of your manuscript with information about your other interests that could influence how they receive and understand your work. The form is designed to be completed electronically and stored electronically. It contains programming that allows appropriate data display. Each author should submit a separate form and is responsible for the accuracy and completeness of the submitted information. The form is in six parts.

1. Identifying information.

2. The work under consideration for publication.

This section asks for information about the work that you have submitted for publication. The time frame for this reporting is that of the work itself, from the initial conception and planning to the present. The requested information is about resources that you received, either directly or indirectly (via your institution), to enable you to complete the work. Checking "No" means that you did the work without receiving any financial support from any third party -- that is, the work was supported by funds from the same institution that pays your salary and that institution did not receive third-party funds with which to pay you. If you or your institution received funds from a third party to support the work, such as a government granting agency, charitable foundation or commercial sponsor, check "Yes".

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This section asks about your financial relationships with entities in the bio-medical arena that could be perceived to influence, or that give the appearance of potentially influencing, what you wrote in the submitted work. You should disclose interactions with ANY entity that could be considered broadly relevant to the work. For example, if your article is about testing an epidermal growth factor receptor (EGFR) antagonist in lung cancer, you should report all associations with entities pursuing diagnostic or therapeutic strategies in cancer in general, not just in the area of EGFR or lung cancer.

Report all sources of revenue paid (or promised to be paid) directly to you or your institution on your behalf over the 36 months prior to submission of the work. This should include all monies from sources with relevance to the submitted work, not just monies from the entity that sponsored the research. Please note that your interactions with the work's sponsor that are outside the submitted work should also be listed here. If there is any question, it is usually better to disclose a relationship than not to do so.

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4. Intellectual Property.

This section asks about patents and copyrights, whether pending, issued, licensed and/or receiving royalties.

5. Relationships not covered above.

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Definitions.

Entity: government agency, foundation, commercial sponsor, academic institution, etc.

Grant: A grant from an entity, generally [but not always] paid to your organization

Personal Fees: Monies paid to you for services rendered, generally honoraria, royalties, or fees for consulting, lectures, speakers bureaus, expert testimony, employment, or other affiliations

Non-Financial Support: Examples include drugs/equipment supplied by the entity, travel paid by the entity, writing assistance, administrative support, etc.

Other: Anything not covered under the previous three boxes

Pending: The patent has been filed but not issued

Issued: The patent has been issued by the agency

Licensed: The patent has been licensed to an entity, whether earning royalties or not

Royalties: Funds are coming in to you or your institution due to your patent

ICMJE Form for Disclosure of Potential Conflicts of Interest

Section 1. Identifying Information

1. Given Name (First Name)
Beyhan

2. Surname (Last Name)
Tuysuz

3. Date
30-October-2015

4. Are you the corresponding author?

☐ Yes ☒ No

Corresponding Author's Name
James R. Lupski

5. Manuscript Title
Molecular etiology of arthrogyrosis in a cohort of families of mostly Turkish origin

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