

#### **Instructions**

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## Identifying information.

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#### 3. Relevant financial activities outside the submitted work.

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## 4. Intellectual Property.

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

# Relationships not covered above.

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earning royalties or not

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

Lupski 1



Section 1. Identifying Inforn	aation			
identifying inform	iation			
Given Name (First Name)  James	2. Surname (Last Nar Lupski	me)		3. Date 26-October-2015
4. Are you the corresponding author?	✓ Yes No			
5. Manuscript Title Molecular etiology of arthrogryposis in	a cohort of families o	of mostly Turkish	origin	
6. Manuscript Identifying Number (if you ki 84457-RG-1	now it)			
Section 2. The Work Under C	onsideration for P	ublication		
Did you or your institution <b>at any time</b> receany aspect of the submitted work (including statistical analysis, etc.)?				
Are there any relevant conflicts of inter-	est? ✓ Yes	No		
If yes, please fill out the appropriate inf Excess rows can be removed by pressin		u have more thar	one entity	press the "ADD" button to add a row.
Name of Institution/Company	Grant? Personal Fees?	Non-Financial Support?	Other?	Comments
NIH/NINDS	<b>✓</b>		U.	54 HG006542
Section 3. Relevant financial	activities outside	the submitted	work.	
Place a check in the appropriate boxes of compensation) with entities as descr clicking the "Add +" box. You should re Are there any relevant conflicts of inter If yes, please fill out the appropriate info	ibed in the instructio port relationships tha est?	ns. Use one line fo	or each enti	ty; add as many lines as you need by
Name of Entity	Grant? Personal Fees?	Non-Financial Support?	Other?	Comments
23 and Me			<b>✓</b> St	tock ownership
Regeneron			<b>√</b> Co	onsultant
asergen			<b>✓</b> St	tock options

Lupski 2



Name of Entity	Grant? Persona	Non-Financial Support?	Other?	Comments	
Miraca Genetics Laboratories		<b>✓</b>		Scientific Advisory Board	
Section 4. Intellectual Propert	y Patents & Co	pyrights			
Do you have any patents, whether plann	ed, pending or issu	ied, broadly releva	nt to the v	vork? ☐ Yes ✓ No	
Section 5. Relationships not c	overed above				
Are there other relationships or activities potentially influencing, what you wrote i			influenced	, or that give the appearance o	f
Yes, the following relationships/conditions/circumstances are present (explain below):					
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At the time of manuscript acceptance, jo On occasion, journals may ask authors to				•	tements.
Section 6. Disclosure Stateme	nt				
Based on the above disclosures, this form below.	n will automatically	generate a disclos	sure stater	nent, which will appear in the b	юх
Dr. Lupski reports grants from NIH/NIND other from Lasergen, non-financial supp	-			_	on,

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Gibbs 1



Section 1.	Identifying Inform	nation	
1. Given Name (Fi Richard	rst Name)	2. Surname (Last Nar Gibbs	3. Date 30-October-2015
4. Are you the cor	responding author?	Yes 🗸 No	Corresponding Author's Name James R. Lupski
5. Manuscript Title Molecular etiolog	e gy of arthrogryposis in	a cohort of families o	f mostly Turkish origin
6. Manuscript lder 84457-RG-1	ntifying Number (if you kr	now it)	
	l		
Section 2.	The Work Under Co	onsideration for P	ublication
any aspect of the s statistical analysis, Are there any rela	ubmitted work (including etc.)? evant conflicts of intere	but not limited to gran	from a third party (government, commercial, private foundation, etc.) for its, data monitoring board, study design, manuscript preparation, No
	out the appropriate info be removed by pressin		u have more than one entity press the "ADD" button to add a row.
Name of Institut		Grant? Personal Fees?	Non-Financial Other? Comments
NIH/NINDS		<b>✓</b>	U54 HG006542
Section 3.	Relevant financial	activities outside	the submitted work.
of compensation	) with entities as descri	ibed in the instructior	e whether you have financial relationships (regardless of amount ns. Use one line for each entity; add as many lines as you need by t were <b>present during the 36 months prior to publication</b> .
•	evant conflicts of intere		No
If yes, please fill o	out the appropriate info	ormation below.	
Name of Entity		Grant? Personal Fees?	Non-Financial Support? Comments
Miraca Genetics Labo	ratories		Interim Chief Scientific Officer

Gibbs 2



Section 4. Intellectual Property Patents & Copyrights				
Do you have any patents, whether planned, pending or issued, broadly relevant to the work? Yes Volume No				
Section 5. Relationships not covered above				
Are there other relationships or activities that readers could perceive to have influenced, or that give the appearance of potentially influencing, what you wrote in the submitted work?				
Yes, the following relationships/conditions/circumstances are present (explain below):				
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Dr. Gibbs reports grants from NIH/NINDS, during the conduct of the study; non-financial support from Miraca Genetics Laboratories, outside the submitted work;.				

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Bayram 1



Section 1. Identifying Inform	nation	
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 mo	stly Turkish origin
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Section 2. The Work Under C	onsideration for Publi	cation
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Section 3. Relevant financial	activities outside the	submitted work.
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Section 4. Intellectual Prope	rty Patents & Copyri	ghts
Do you have any patents, whether plan	nned, pending or issued, b	roadly relevant to the work? Yes V No

Bayram 2



Section 5.	
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Yes, the followi	ng relationships/conditions/circumstances are present (explain below):
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	uscript acceptance, journals will ask authors to confirm and, if necessary, update their disclosure statements als may ask authors to disclose further information about reported relationships.
Section 6.	Disclosure Statement
Based on the above below.	e disclosures, this form will automatically generate a disclosure statement, which will appear in the box
Dr. Bayram has not	thing to disclose.

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Karaca 1



Section 1. Identifying Inform	nation	
1. Given Name (First Name) Ender	2. Surname (Last Name) Karaca	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 mo	stly Turkish origin
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Karaca 2



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Dr. Karaca has nothing to disclose.

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Coban Akdemir 1



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4. Are you the corresponding author?	☐ Yes ✓ No	Corresponding Author's Name James R. Lupski
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Do you have any patents, whether plan		

Coban Akdemir 2



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

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

Ozdamar Yilmaz 1



Section 1. Identifying Infor	mation	
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 i	n a cohort of families of mo	stly Turkish origin
6. Manuscript Identifying Number (if you l 84457-RG-1	know it)	
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Section 2. The Work Under (	Consideration for Public	cation
	ng but not limited to grants, da	a third party (government, commercial, private foundation, etc.) for ata monitoring board, study design, manuscript preparation,
Section 3. Relevant financia	l activities outside the s	submitted work.
of compensation) with entities as desc	cribed in the instructions. Us eport relationships that wer	ether you have financial relationships (regardless of amount se one line for each entity; add as many lines as you need by re <b>present during the 36 months prior to publication</b> .
Section 4. Intellectual Property		
Intellectual Prope	erty Patents & Copyric	gnts ———
Do you have any patents, whether pla	nned, pending or issued, br	roadly relevant to the work? Yes V No

Ozdamar Yilmaz 2



Section 5. Relationships not sovered above
Relationships not covered above
Are there other relationships or activities that readers could perceive to have influenced, or that give the appearance of potentially influencing, what you wrote in the submitted work?
Yes, the following relationships/conditions/circumstances are present (explain below):
✓ No other relationships/conditions/circumstances that present a potential conflict of interest
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Dr. Ozdamar Yilmaz has nothing to disclose.

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Ozdamar Yilmaz 3



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

Akay Tayfun 1



Section 1. Identifying Inform	nation	
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 James R. Lupski
5. Manuscript Title Molecular etiology of arthrogryposis in	a cohort of families of mo	stly Turkish origin
6. Manuscript Identifying Number (if you k	now it)	
Section 2. The Work Under C	onsideration for Public	cation
	g but not limited to grants, da	a third party (government, commercial, private foundation, etc.) for ata monitoring board, study design, manuscript preparation,
Section 3. Relevant financial	activities outside the	submitted work.
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Section 4. Intellectual Prope	rty Patents & Copyri	ghts
Do you have any patents, whether plan	nned, pending or issued, br	roadly relevant to the work? Yes V No

Akay Tayfun 2



Section 5.	
Section 5.	Relationships not covered above
	elationships or activities that readers could perceive to have influenced, or that give the appearance of encing, what you wrote in the submitted work?
Yes, the follow	wing relationships/conditions/circumstances are present (explain below):
✓ No other rela	tionships/conditions/circumstances that present a potential conflict of interest
	anuscript acceptance, journals will ask authors to confirm and, if necessary, update their disclosure statements rnals may ask authors to disclose further information about reported relationships.
Section 6.	Disclosure Statement
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Dr. Akay Tayfun	has nothing to disclose.

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Akay Tayfun 3



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

Aydin 1



Section 1.	Identifying Inform	ation	
1. Given Name (First Hatip	st Name)	2. Surname (Last Name) Aydin	3. Date 30-October-2015
4. Are you the corre	esponding author?	Yes ✓ No	Corresponding Author's Name James R. Lupski
5. Manuscript Title Molecular etiolog	y of arthrogryposis in	a cohort of families of mo	ostly Turkish origin
6. Manuscript Iden 84457-RG-1	tifying Number (if you kn	ow it)	
			_
Section 2.	The Work Under Co	onsideration for Publi	cation
any aspect of the su statistical analysis, e	ıbmitted work (including	but not limited to grants, d	n a third party (government, commercial, private foundation, etc.) for ata monitoring board, study design, manuscript preparation,
Section 3.	Relevant financial	activities outside the	submitted work.
of compensation) clicking the "Add	with entities as descri	bed in the instructions. Uport relationships that we	nether you have financial relationships (regardless of amount se one line for each entity; add as many lines as you need by tre present during the 36 months prior to publication.
Section 4.	Intellectual Proper	ty Patents & Copyri	ghts
			roadly relevant to the work? Yes V No

Aydin 2



Section 5.				
	Relationships not covered above			
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Yes, the follo	wing relationships/conditions/circumstances are present (explain below):			
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Section 6.	Disclosure Statement			
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Torun 1



Section 1.	Identifying Inform	nation	
1. Given Name (Fir Deniz	rst Name)	2. Surname (Last Name) Torun	3. Date 30-October-2015
4. Are you the cor	responding author?	Yes ✓ No	Corresponding Author's Name James R. Lupski
5. Manuscript Title Molecular etiolog		a cohort of families of mo	stly Turkish origin
6. Manuscript Ider 84457-RG-1	ntifying Number (if you kr	now it)	
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of compensation clicking the "Add	) with entities as descri	ibed in the instructions. Us port relationships that we	nether you have financial relationships (regardless of amount se one line for each entity; add as many lines as you need by tree present during the 36 months prior to publication.
Section 4.	Intellectual Proper	rty Patents & Copyri	ghts
Do you have any	patents, whether plan	ned, pending or issued, br	roadly relevant to the work? Yes V No

Torun 2



Section 5. Relationships not covered above
helationships not covered above
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or other affiliations patent

Tug Bozdogan 1



Section 1. Identifying Inform	nation	
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
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Place a check in the appropriate boxes in the appropriate boxes in the appropriate boxes in the compensation.	in the table to indicate wh ibed in the instructions. U port relationships that we	nether you have financial relationships (regardless of amount se one line for each entity; add as many lines as you need by re present during the 36 months prior to publication.
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Do you have any patents, whether plan	ned, pending or issued, b	roadly relevant to the work? Yes Vo

Tug Bozdogan 2



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Tug Bozdogan 3



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Gezdirici 1



Section 1. Identifying Inform	nation	
1. Given Name (First Name) Alper	2. Surname (Last Name) Gezdirici	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 ir	a cohort of families of mo	stly Turkish origin
6. Manuscript Identifying Number (if you k 84457-RG-1	now it)	
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of compensation) with entities as desc	ribed in the instructions. Use port relationships that we	ether you have financial relationships (regardless of amount se one line for each entity; add as many lines as you need by re present during the 36 months prior to publication.
Section 4. Intellectual Proper		
Intellectual Prope	rty Patents & Copyri	ghts
Do you have any patents, whether plan	nned, pending or issued, br	oadly relevant to the work? Yes V No

Gezdirici 2



Section 5. Relationships not covered above
Are there other relationships or activities that readers could perceive to have influenced, or that give the appearance of potentially influencing, what you wrote in the submitted work?
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Dr. Gezdirici has nothing to disclose.

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Gezdirici 3



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

Isikay 1



Section 1. Identifying Inform	mation	
1. Given Name (First Name) Sedat	2. Surname (Last Name) Isikay	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 ir	n a cohort of families of mo	stly Turkish origin
6. Manuscript Identifying Number (if you k 84457-RG-1	now it)	
		_
Section 2. The Work Under C	Consideration for Public	cation
	g but not limited to grants, da	a third party (government, commercial, private foundation, etc.) for ita monitoring board, study design, manuscript preparation,
Section 3. Polovent Guardia	l activities outside the s	ماريد المعدد ما المعادد
Place a check in the appropriate boxes of compensation) with entities as desc	in the table to indicate who ribed in the instructions. Us eport relationships that wer	ether you have financial relationships (regardless of amount se one line for each entity; add as many lines as you need by re present during the 36 months prior to publication.
Section 4. Intellectual Prope	erty Patents & Copyric	ahts
Do you have any patents, whether plan		

Isikay 2



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Are there other relationships or activities that readers could perceive to have influenced, or that give the appearance of potentially influencing, what you wrote in the submitted work?
Yes, the following relationships/conditions/circumstances are present (explain below):  No other relationships/conditions/circumstances that present a potential conflict of interest
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Section 6. Disclosure Statement
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Dr. Isikay has nothing to disclose.

## **Evaluation and Feedback**

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Isikay 3



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

Atik 1



Section 1.	Identifying Inform	nation	
1. Given Name (Fi Mehmed	. , ,	2. Surname (Last Name) Atik	3. Date 30-October-2015
4. Are you the cor	responding author?	Yes ✓ No	Corresponding Author's Name  James R. Lupski
5. Manuscript Title Molecular etiolo		a cohort of families of mo	stly Turkish origin
6. Manuscript Ide	ntifying Number (if you kr	now it)	
			_
Section 2.	The Work Under C	onsideration for Public	cation
any aspect of the s statistical analysis,	ubmitted work (including	g but not limited to grants, da	a third party (government, commercial, private foundation, etc.) for ata monitoring board, study design, manuscript preparation,
Section 3.	Relevant financial	activities outside the s	submitted work.
of compensation clicking the "Add	n) with entities as descri	ibed in the instructions. Us port relationships that wer	ether you have financial relationships (regardless of amount se one line for each entity; add as many lines as you need by re present during the 36 months prior to publication.
Section 4.	Intellectual Proper	rty Patents & Copyrig	ahts
Do you have any			roadly relevant to the work? Yes V No

Atik 2



Section 5. Relationships not covered above
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Dr. Atik has nothing to disclose.

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Gambin 1



Section 1.	dentifying Informa	ation		
1. Given Name (First l	Name)	2. Surname (Last Name) Gambin		3. Date 30-October-2015
4. Are you the corres	ponding author?	Yes 🗸 No	Corresponding Author's Nar James R. Lupski	me
5. Manuscript Title Molecular etiology	of arthrogryposis in a	a cohort of families of m	ostly Turkish origin	
6. Manuscript Identif 84457-RG-1	ying Number (if you kno	ow it)		
Section 2. T	he Work Under Co	nsideration for Pub	lication	
any aspect of the subs statistical analysis, etc	mitted work (including	but not limited to grants, o	m a third party (government, cor data monitoring board, study de	mmercial, private foundation, etc.) for sign, manuscript preparation,
Section 3. R	elevant financial a	activities outside the	submitted work.	
of compensation) w clicking the "Add +'	vith entities as describ	oed in the instructions. I ort relationships that w		ationships (regardless of amount dd as many lines as you need by nonths prior to publication.
Section 4.	ntellectual Propert	ty Patents & Copyr	ights	
Do you have any pa	itents, whether plann	ed, pending or issued, l	proadly relevant to the work?	Yes 🗸 No

Gambin 2



Section 5. Relationships not covered above
Relationships not covered above
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Harel 1



Section 1. Identifying Inform	nation		
1. Given Name (First Name) Tamar	2. Surname (Last Name) Harel	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 mo	stly Turkish origin	
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Section 2. The Work Under Co	onsideration for Publi	cation	
	but not limited to grants, da	a third party (government, commercial, private foundation, e ata monitoring board, study design, manuscript preparation,	tc.) for
Section 3. Relevant financial	activities outside the	submitted work.	
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Section 4. Intellectual Proper	ety Datante & Comuni	nhtc.	
intellectual Propel	rty Patents & Copyri	gnits ———	
Do you have any patents, whether plan	ned, pending or issued, b	roadly relevant to the work? Yes V No	

Harel 2



Section 5.	Balatian diamentary and allower				
	Relationships not covered above				
	elationships or activities that readers could perceive to have influenced, or that give the appearance of encing, what you wrote in the submitted work?				
Yes, the follow	wing relationships/conditions/circumstances are present (explain below):				
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Section 6.	Disclosure Statement				
Based on the abo below.	ove disclosures, this form will automatically generate a disclosure statement, which will appear in the box				
Dr. Harel has not	thing to disclose.				

# **Evaluation and Feedback**

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El-Hattab 1



Section 1. Identifying Inform	nation	
1. Given Name (First Name) Ayman	2. Surname (Last Name) El-Hattab	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 mo	stly Turkish origin
6. Manuscript Identifying Number (if you ki 84457-RG-1	now it)	
Section 2. The Work Under C	onsideration for Public	cation
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Section 4. Intellectual Prope		
Intellectual Prope	rty Patents & Copyri	ghts ————
Do you have any patents, whether plan	ned, pending or issued, br	oadly relevant to the work? Yes V No

El-Hattab 2



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Relationships not covered above
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Dr. El-Hattab has nothing to disclose.

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

Charng 1



Section 1. Identifying Inform	nation	
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 mo	stly Turkish origin
6. Manuscript Identifying Number (if you ki 84457-RG-1	now it)	
Section 2. The Work Under C	onsideration for Publi	cation
	g but not limited to grants, da	a third party (government, commercial, private foundation, etc.) for ata monitoring board, study design, manuscript preparation,
Section 3. Relevant financial	activities outside the	submitted work.
of compensation) with entities as descr	ibed in the instructions. Us port relationships that we	ether you have financial relationships (regardless of amount se one line for each entity; add as many lines as you need by re <b>present during the 36 months prior to publication</b> .
Section 4. Intellectual Prope	rty Patents & Copyri	ghts
Do you have any patents, whether plan	ned, pending or issued, b	roadly relevant to the work? Yes V No

Charng 2



Section 5. Relationships not covered above
Relationships not covered above
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Dr. Charng has nothing to disclose.

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Charng 3



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Pehlivan 1



Section 1.	Identifying Inform	nation	
1. Given Name (Fir Davut	st Name)	2. Surname (Last Name) Pehlivan	3. Date 30-October-2015
4. Are you the corr	esponding author?	Yes ✓ No	Corresponding Author's Name James R. Lupski
5. Manuscript Title Molecular etiolog		a cohort of families of mo	stly Turkish origin
6. Manuscript Iden 84457-RG-1	itifying Number (if you kn	now it)	
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any aspect of the su statistical analysis, o	ubmitted work (including	but not limited to grants, da	a third party (government, commercial, private foundation, etc.) for ita monitoring board, study design, manuscript preparation,
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Section 4.	Intellectual Proper	ty Patents & Copyri	ghts
Do you have any	patents, whether plan	ned, pending or issued, br	oadly relevant to the work? Yes V No

Pehlivan 2



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Jhangiani 1



Section 1. Identifying Inform	mation	
1. Given Name (First Name) Shalini	2. Surname (Last Name) Jhangiani	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 ir	n a cohort of families of mo	stly Turkish origin
6. Manuscript Identifying Number (if you k 84457-RG-1	now it)	
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	g but not limited to grants, da	a third party (government, commercial, private foundation, etc.) for ta monitoring board, study design, manuscript preparation,
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Intellectual Prope	rty Patents & Copyric	ghts ———
Do you have any patents, whether plan	nned, pending or issued, br	oadly relevant to the work? Yes V No

Jhangiani 2



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Muzny 1



Section 1.	Identifying Inform	nation	
1. Given Name (Fii Donna	rst Name)	2. Surname (Last Name) Muzny	3. Date 30-October-2015
4. Are you the cor	responding author?	☐ Yes ✓ No	Corresponding Author's Name James R. Lupski
5. Manuscript Title Molecular etiolo		a cohort of families of mo	stly Turkish origin
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Section 4.	Intellectual Proper	rty Patents & Copyric	ghts
Do you have any	patents, whether plan	ned, pending or issued, br	roadly relevant to the work? Yes V No

Muzny 2



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Karaman 1



Section 1. Identifying Inform	nation	
1. Given Name (First Name) Ali	2. Surname (Last Name) Karaman	3. Date 30-October-2015
4. Are you the corresponding author?	Yes Vo	Corresponding Author's Name James R. Lupski
5. Manuscript Title Molecular etiology of arthrogryposis in	a cohort of families of mo	stly Turkish origin
6. Manuscript Identifying Number (if you k 84457-RG-1	now it)	
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Intellectual Prope	rty Patents & Copyri	gnts
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Karaman 2



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Celik 1



Section 1.	Identifying Inform	ation		
1. Given Name (First Tamer	: Name)	2. Surname (Last Name) Celik		3. Date 30-October-2015
4. Are you the corre	sponding author?	Yes ✓ No	Corresponding Author's Nar James R. Lupski	me
5. Manuscript Title Molecular etiology	of arthrogryposis in a	a cohort of families of mo	ostly Turkish origin	
6. Manuscript Identi 84457-RG-1	ifying Number (if you kn	ow it)		
			_	
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any aspect of the sub statistical analysis, et	omitted work (including	but not limited to grants, d	n a third party (government, cor ata monitoring board, study de	mmercial, private foundation, etc.) for sign, manuscript preparation,
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of compensation) of clicking the "Add +	with entities as descri	bed in the instructions. Uport relationships that we	se one line for each entity; a	ationships (regardless of amount dd as many lines as you need by conths prior to publication.
Section 4.	ntellectual Proper	ty Patents & Copyri	ghts	
Do you have any p	atents, whether planr	ned, pending or issued, b	roadly relevant to the work?	☐ Yes 🗸 No

Celik 2



Section 5. Relationships not sovered above
Relationships not covered above
Are there other relationships or activities that readers could perceive to have influenced, or that give the appearance of potentially influencing, what you wrote in the submitted work?
Yes, the following relationships/conditions/circumstances are present (explain below):
✓ No other relationships/conditions/circumstances that present a potential conflict of interest
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
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Dr. Celik has nothing to disclose.

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Celik 3



#### **Instructions**

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

Ozalp Yuregir 1



Section 1.	Identifying Inform	nation	
1. Given Name (Fii Ozge	rst Name)	2. Surname (Last Name) Ozalp Yuregir	3. Date 30-October-2015
4. Are you the cor	responding author?	Yes ✓ No	Corresponding Author's Name James R. Lupski
5. Manuscript Title Molecular etiolo		a cohort of families of mo	stly Turkish origin
6. Manuscript Ider 84457-RG-1	ntifying Number (if you kr	now it)	
Section 2.	The Work Under C	onsideration for Public	cation
any aspect of the s statistical analysis,	ubmitted work (including	g but not limited to grants, da	a third party (government, commercial, private foundation, etc.) for ata monitoring board, study design, manuscript preparation,
Section 3.	Relevant financial	activities outside the s	submitted work.
of compensation clicking the "Add	) with entities as descri	ibed in the instructions. Us port relationships that wer	ether you have financial relationships (regardless of amount se one line for each entity; add as many lines as you need by re <b>present during the 36 months prior to publication</b> .
Section 4.	Intellectual Prope	rty Patents & Copyric	ghts
Do you have any	patents, whether plan	ned, pending or issued, br	roadly relevant to the work? Yes V No

Ozalp Yuregir 2



Section 5. Relationships not covered above
Relationships not covered above
Are there other relationships or activities that readers could perceive to have influenced, or that give the appearance of potentially influencing, what you wrote in the submitted work?
Yes, the following relationships/conditions/circumstances are present (explain below):
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Ozalp Yuregir 3



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

Yildirim 1



Section 1. Identifying Inform	nation	
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 mo	stly Turkish origin
6. Manuscript Identifying Number (if you kr 84457-RG-1	now it)	
		_
Section 2. The Work Under Co	onsideration for Publi	cation
	but not limited to grants, da	n a third party (government, commercial, private foundation, etc.) for ata monitoring board, study design, manuscript preparation,
Section 3. Relevant financial	activities outside the	submitted work.
of compensation) with entities as descri	ibed in the instructions. U port relationships that we	nether you have financial relationships (regardless of amount se one line for each entity; add as many lines as you need by re <b>present during the 36 months prior to publication</b> .
Section 4. Intellectual Proper	utu. Datanta 9 Canusi	white
intellectual Propel	rty Patents & Copyri	gnts ———
Do you have any patents, whether plan	ned, pending or issued, b	roadly relevant to the work? Yes V No

Yildirim 2



Section 5. Relationships not covered above
Relationships not covered above
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## Relationships not covered above.

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

patent

Bayhan 1



Section 1.	Identifying Inform	nation		
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 Nam James R. Lupski	ne
5. Manuscript Title Molecular etiolog	yy of arthrogryposis in	a cohort of families of mo	ostly Turkish origin	
6. Manuscript Iden 84457-RG-1	tifying Number (if you kn	now it)		
Section 2.	The Work Under Co	onsideration for Publi	cation	
Did you or your institution <b>at any time</b> 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				
Section 3.	Relevant financial	activities outside the	submitted work.	
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Section 4.	Intellectual Proper	ty Patents & Copyri	ghts	
Do you have any patents, whether planned, pending or issued, broadly relevant to the work? Yes Vo				

Bayhan 2



Section 5. Relationships not sovered above
Relationships not covered above
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**Royalties:** Funds are coming in to you or your institution due to your patent

Boerwinkle 1



Section 1.	Identifying Inform	ation		
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 Nan James R. Lupski	ne
5. Manuscript Title Molecular etiolog		a cohort of families of mo	ostly Turkish origin	
6. Manuscript Ider 84457-RG-1	6. Manuscript Identifying Number (if you know it) 84457-RG-1			
Section 2.	The Work Under Co	onsideration for Publi	ication	
Did you or your institution <b>at any time</b> 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  Yes  No				
Section 3.	Relevant financial	activities outside the	submitted work	
of compensation clicking the "Add	the appropriate boxes i ) with entities as descri	n the table to indicate wh bed in the instructions. U port relationships that we	nether you have financial rela	ationships (regardless of amount dd as many lines as you need by onths prior to publication.
Section 4.	Intellectual Proper	ty Patents & Copyri	ahts	
Do you have any			proadly relevant to the work?	☐ Yes 🗸 No

Boerwinkle 2



Section 5. Polationships not sovered above
Relationships not covered above
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Yes, the following relationships/conditions/circumstances are present (explain below):
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Boerwinkle 3



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Elcioglu 1



Section 1.	Identifying Inform	nation		
1. Given Name (First Name) Nursel		2. Surname (Last Name) Elcioglu	3. Date 30-October-2015	
4. Are you the corresponding author?		Yes ✓ No	Corresponding Author's Name James R. Lupski	
5. Manuscript Title Molecular etiolo		a cohort of families of mo	stly Turkish origin	
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Section 3.	Relevant financial	activities outside the s	submitted work.	
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Elcioglu 2



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Dr. Elcioglu has nothing to disclose.			

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Elcioglu 3



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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".

#### 3. Relevant financial activities outside the submitted work.

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.

For grants you have received for work outside the submitted work, you should disclose support ONLY from entities that could be perceived to be affected financially by the published work, such as drug companies, or foundations supported by entities that could be perceived to have a financial stake in the outcome. Public funding sources, such as government agencies, charitable foundations or academic institutions, need not be disclosed. For example, if a government agency sponsored a study in which you have been involved and drugs were provided by a pharmaceutical company, you need only list the pharmaceutical company.

## 4. Intellectual Property.

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

## Relationships not covered above.

Use this section to report other relationships or activities that readers could perceive to have influenced, or that give the appearance of potentially influencing, what you wrote in the submitted work.

#### 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

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Section 1.	Identifying Inform	nation			
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 etiolo		a cohort of families of mo	stly Turkish origin		
6. Manuscript Ider 84457-RG-1	6. Manuscript Identifying Number (if you know it) 84457-RG-1				
Section 2.	The Work Under Co	onsideration for Publi	cation		
Did you or your institution <b>at any time</b> 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 V					
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 <b>present during the 36 months prior to publication</b> .  Are there any relevant conflicts of interest? Yes Vo					
Section 4.	Intellectual Proper	ty Patents & Copyri	ghts		
Do you have any patents, whether planned, pending or issued, broadly relevant to the work? Yes Vo					

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Section 5. Relationships not covered above				
Are there other relationships or activities that readers could perceive to have influenced, or that give the appearance of potentially influencing, what you wrote in the submitted work?				
Yes, the following relationships/conditions/circumstances are present (explain below):				
✓ No other relationships/conditions/circumstances that present a potential conflict of interest				
At the time of manuscript acceptance, journals will ask authors to confirm and, if necessary, update their disclosure statement 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. Tuysuz has nothing to disclose.				

#### **Evaluation and Feedback**

Please visit <a href="http://www.icmje.org/cgi-bin/feedback">http://www.icmje.org/cgi-bin/feedback</a> to provide feedback on your experience with completing this form.

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