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ESPS PEER-REVIEW REPORT

Name of journal: World Journal of Medical Genetics

ESPS manuscript NO: 15311

Title: Clinical applications of high-throughput genetic diagnosis in retinal dystrophies: present challenges and future directions

Reviewer's code: 00631992

Reviewer's country: Italy

Science editor: Xue-Mei Gong

Date sent for review: 2014-11-24 08:27

Date reviewed: 2014-12-01 03:41

CLASSIFICATION	LANGUAGE EVALUATION	SCIENTIFIC MISCONDUCT	CONCLUSION
<input type="checkbox"/> Grade A: Excellent	<input checked="" type="checkbox"/> Grade A: Priority publishing	PubMed Search:	<input checked="" type="checkbox"/> Accept
<input checked="" type="checkbox"/> Grade B: Very good	<input type="checkbox"/> Grade B: Minor language polishing	<input type="checkbox"/> The same title	<input type="checkbox"/> High priority for publication
<input type="checkbox"/> Grade C: Good		<input type="checkbox"/> Duplicate publication	
<input type="checkbox"/> Grade D: Fair	<input type="checkbox"/> Grade C: A great deal of language polishing	<input type="checkbox"/> Plagiarism	<input type="checkbox"/> Rejection
<input type="checkbox"/> Grade E: Poor	<input type="checkbox"/> Grade D: Rejected	<input checked="" type="checkbox"/> No	<input type="checkbox"/> Minor revision
		BPG Search:	<input type="checkbox"/> Major revision
		<input type="checkbox"/> The same title	
		<input type="checkbox"/> Duplicate publication	
		<input type="checkbox"/> Plagiarism	
		<input checked="" type="checkbox"/> No	

COMMENTS TO AUTHORS

The Authors have focused on the latest advances in the molecular diagnosis of inherited retinal disorders using Next Generation Sequencing methodologies, highlighting their actual strengths and weaknesses, and providing arguments to support their possible role for the routine molecular diagnosis of hereditary diseases in the near future. The manuscript is well written and has a good readability. It offers a brief but fairly complete view of the subject matter, which is very timely and important. Only two little remarks: 1. The abstract lacks of the last words (diagnosis in the future). 2. From a formal point of view it would be better to use a single definition for the abbreviations used. In particular, the abbreviation IRDs, which is central to the economy of the manuscript, is defined as "Inherited retinal dystrophies" in keywords, and as "Inherited retinal degenerations" in the introduction.



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ESPS PEER-REVIEW REPORT

Name of journal: World Journal of Medical Genetics

ESPS manuscript NO: 15311

Title: Clinical applications of high-throughput genetic diagnosis in retinal dystrophies: present challenges and future directions

Reviewer's code: 00505250

Reviewer's country: United Kingdom

Science editor: Xue-Mei Gong

Date sent for review: 2014-11-24 08:27

Date reviewed: 2014-12-04 22:32

CLASSIFICATION	LANGUAGE EVALUATION	SCIENTIFIC MISCONDUCT	CONCLUSION
<input type="checkbox"/> Grade A: Excellent	<input type="checkbox"/> Grade A: Priority publishing	PubMed Search:	<input type="checkbox"/> Accept
<input type="checkbox"/> Grade B: Very good	<input checked="" type="checkbox"/> Grade B: Minor language polishing	<input type="checkbox"/> The same title	<input type="checkbox"/> High priority for publication
<input checked="" type="checkbox"/> Grade C: Good		<input type="checkbox"/> Duplicate publication	
<input type="checkbox"/> Grade D: Fair	<input type="checkbox"/> Grade C: A great deal of language polishing	<input type="checkbox"/> Plagiarism	<input type="checkbox"/> Rejection
<input type="checkbox"/> Grade E: Poor		<input checked="" type="checkbox"/> No	<input checked="" type="checkbox"/> Minor revision
	<input type="checkbox"/> Grade D: Rejected	BPG Search:	<input type="checkbox"/> Major revision
		<input type="checkbox"/> The same title	
		<input type="checkbox"/> Duplicate publication	
		<input type="checkbox"/> Plagiarism	
		<input checked="" type="checkbox"/> No	

COMMENTS TO AUTHORS

This was a very interesting and enjoyable read from a clinician's point of view. However, I am not an expert of genetics and would highly recommend a review by an ophthalmic genetics expert as I am unable to comment on the specific genetics. However, from a clinician's point of view, this is a very interesting paper.