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ESPS Peer-review Report

Name of Journal: World Journal of Clinical Cases

ESPS Manuscript NO: 10604

Title: Rare large homozygous CFTR gene deletion in an Iranian patient with cystic fibrosis

Reviewer code: 00403513

Science editor: Xiu-Xia Song

Date sent for review: 2014-04-09 19:17

Date reviewed: 2014-04-24 19:50

CLASSIFICATION	LANGUAGE EVALUATION	RECOMMENDATION	CONCLUSION
<input type="checkbox"/> Grade A (Excellent)	<input type="checkbox"/> Grade A: Priority Publishing	Google Search:	<input type="checkbox"/> Accept
<input type="checkbox"/> Grade B (Very good)	<input checked="" type="checkbox"/> Grade B: minor language polishing	<input type="checkbox"/> Existed	<input type="checkbox"/> High priority for publication
<input checked="" type="checkbox"/> Grade C (Good)	<input type="checkbox"/> Grade C: a great deal of language polishing	<input type="checkbox"/> No records	<input type="checkbox"/> Rejection
<input type="checkbox"/> Grade D (Fair)		BPG Search:	<input type="checkbox"/> Rejection
<input type="checkbox"/> Grade E (Poor)	<input type="checkbox"/> Grade D: rejected	<input type="checkbox"/> Existed	<input checked="" type="checkbox"/> Minor revision
		<input type="checkbox"/> No records	<input type="checkbox"/> Major revision

COMMENTS TO AUTHORS

The manuscript "Rare large homozygous CFTR gene deletion in an Iranian patient with Cystic Fibrosis" by Farjadian and colleagues reports a patient with homozygous exon 4-10 CFTR gene deletion mutation. Overall, this manuscript is well written and suitable as a case-report. Some minor issues might be considered previous to publication

- 1)General: Please proof-read English grammar .
- 2)The reported patient has parents with first-degree consanguinity. Are there any further symptoms which might not be related to CF ? Could a syndrome i.e. due to multiple gene mutations be ruled out ?
- 3) Did the heterozygous parents/sister/brother show abnormal sweat tests or any sign of mild CF disease?
- 4) Was genetic counseling performed for the family and the patient?



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ESPS Peer-review Report

Name of Journal: World Journal of Clinical Cases

ESPS Manuscript NO: 10604

Title: Rare large homozygous CFTR gene deletion in an Iranian patient with cystic fibrosis

Reviewer code: 00029421

Science editor: Xiu-Xia Song

Date sent for review: 2014-04-09 19:17

Date reviewed: 2014-04-25 19:40

CLASSIFICATION	LANGUAGE EVALUATION	RECOMMENDATION	CONCLUSION
<input type="checkbox"/> Grade A (Excellent)	<input type="checkbox"/> Grade A: Priority Publishing	Google Search:	<input type="checkbox"/> Accept
<input type="checkbox"/> Grade B (Very good)	<input checked="" type="checkbox"/> Grade B: minor language polishing	<input type="checkbox"/> Existed	<input type="checkbox"/> High priority for publication
<input checked="" type="checkbox"/> Grade C (Good)	<input type="checkbox"/> Grade C: a great deal of language polishing	<input type="checkbox"/> No records	<input type="checkbox"/> Rejection
<input type="checkbox"/> Grade D (Fair)		BPG Search:	<input type="checkbox"/> Rejection
<input type="checkbox"/> Grade E (Poor)	<input type="checkbox"/> Grade D: rejected	<input type="checkbox"/> Existed	<input checked="" type="checkbox"/> Minor revision
		<input type="checkbox"/> No records	<input type="checkbox"/> Major revision

COMMENTS TO AUTHORS

Straightforward focussed and relatively well written Ms describing a patient with CF among a population where this condition is not common. A rare but not unprecedented large gene deletion was identified - the unusual nature of the case is partially related to the fact that this deletion was homozygous due to parental consanguinity which resulted in unusual results (no delta F508 allele) on initial testing. Minor text changes needed It might be interesting to know what frequency and the spectrum of CF mutations are in this population.