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ESPS PEER REVIEW REPORT

Name of journal: World Journal of Clinical Cases

ESPS manuscript NO: 10929

Title: Concomitant Achondroplasia and Chiari II malformation: a double-hit at the cervicomedullary junction.

Reviewer code: 00542914

Science editor: Ling-Ling Wen

Date sent for review: 2014-04-27 22:47

Date reviewed: 2014-06-10 21:12

CLASSIFICATION	LANGUAGE EVALUATION	RECOMMENDATION	CONCLUSION
<input type="checkbox"/> Grade A: Excellent	<input checked="" type="checkbox"/> Grade A: Priority publishing	Google 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"/> Existing	<input type="checkbox"/> High priority for publication
<input 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	<input type="checkbox"/> Grade D: Rejected	BPG Search:	<input type="checkbox"/> Minor revision
<input type="checkbox"/> Grade E: Poor		<input type="checkbox"/> Existing	<input type="checkbox"/> Major revision
		<input type="checkbox"/> No records	

COMMENTS TO AUTHORS

The ms of Awad et al describes a neonate case with concomitant achondroplasia and Chiari II malformation (CMII). According to a literature review performed by the authors this is the first case report of a patient with this combination. This is definitely an interesting case that is thoroughly documented. Even though I am not an expert in the interpretation of MRI images everything seems to be very well described. Apparently, the genetic cause for CMII has not yet been identified but animal models and candidate genes are nicely discussed. However, with regard to the potential link between the two conditions I would find it interesting to show or at least mention the result of the FGFR3 mutation analysis. I know that 99% of the achondroplasia cases are caused by the Gly380Arg substitution but a lot more very rare mutations were identified over the last years. Maybe the knowledge of the mutation could give some insight into the underlying pathomechanism especially as some of the very rare mutations in FGFR3 were also associated with cranial malformations, like e.g. Crouzon syndrome, Muenke syndrome and others.