



SDI Review Form 1.6

Journal Name:	<u>British Journal of Medicine and Medical Research</u>
Manuscript Number:	2015_BJMMR_18367
Title of the Manuscript:	Majewski syndrome, Short Rib Polydactyly Syndrome (SRPS) Type II - A rare case report, with review of literature
Type of the Article	Case Study

General guideline for Peer Review process:

This journal's peer review policy states that **NO** manuscript should be rejected only on the basis of '**lack of Novelty**', provided the manuscript is scientifically robust and technically sound.

To know the complete guideline for Peer Review process, reviewers are requested to visit this link:

(<http://www.sciencedomain.org/page.php?id=sdi-general-editorial-policy#Peer-Review-Guideline>)



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PART 1: Review Comments

	Reviewer's comment	Author's comment (if agreed with reviewer, correct the manuscript and highlight that part in the manuscript. It is mandatory that authors should write his/her feedback here)
Compulsory REVISION comments	This case report does not say anything in addition to what is known on this malformation 1. Was there any unique findings in this case that merits publication and if so then those findings should be stressed/highlighted 2. Review also is very short as so discussion 3. Many things at molecular level is possible with lethal skeletal dysplasia including NGS panel for lethal skeletal dysplasia which could be used not only for diagnosis and screening but also early prenatal diagnosis from CVS samples or even PGD (in case IVF is indicated or several sibs affected) 4. D/D with Mohr syndrome 5. Discussion should cover molecular mechanism including molecular classification	1. Unique findings in this case which have either not been reported or very rarely reported in the past are- short small intestine, hypoplastic epiglottis and larynx, hepatic fibrosis and atrial septal defect. This case report was prepared because of its extreme rarity and limited literature which deals with all the features of Majewski syndrome in detail. 2. Case discussion has been made more extensive. 3. Due to limitations in diagnostic modalities available at our institution, the investigations mentioned could not be performed. 4. Mohr syndrome has been added in the differential diagnosis. 5. Molecular mechanisms have been added in the case discussion.
Minor REVISION comments	1. Case description as well as discussion should be continuous without interruption by figures or tables Figure & tables may be given at the end	1. Figures and Tables have been placed at the end after the references



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	2. Figure I- X Ray Infantogram: polydactyly not appreciable 3. Figure VI- Both hands of the same: in fact should be left hand (no need to use same as single case report) Figure VII- Both feet of the same: in fact should be left foot (no need to use same as single case report) Figure XII- Photomicrograph of Tibia & Costochondral junction: as photograph is from single section this should be from either costochondral junction or tibia (cannot be both) 4. Too many figures: many can be omitted viz., 1 or 2; 3; 8-11 5. Table subheads may be limited to type I to IV and present case	2. Figure I - X Ray Infantogram did not highlight the polydactyly very well and that is why it was followed by a CT scan. 3. Corrections have been made in the figure legends. 4. Figure I –Xray removed because the features were better highlighted in the CT scan. Some figures have been clubbed together as one. Total number of figures in the revised manuscript is 5. 5. Table has been modified to include only the 4 types of SRPS and the present case.
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<u>Optional/General</u> comments	Although fetal autopsy is carried out after radiological investigation however it is adviseable to provide details of clinical description including internal at the beginning and thereafter investigatory findings viz., radiological, histopathological or molecular	We have broken down the case presentation into various subheadings i.e. Pregnancy history, Prenatal USG findings, Birth and Perinatal history, External examination, Postnatal imaging and Autopsy findings- external and internal including the gross and microscopic findings. The case description has been drafted in a step-wise manner to detail the protocol that was followed by us.
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