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Diagnosis in Infants and Children With Osteogenesis Imperfecta —author response

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We are grateful for Drs. Shelmerdine, Arthurs and Calder's interest in our article "Fractures at Diagnosis in Infants and Children with Osteogenesis Imperfecta".¹ We would like to address the concerns raised by the authors in their letter² as we believe that they arise from a misunderstanding of the intent and methodology of our study.

First, Dr. Shelmerdine and colleagues criticize our study as both not having a control group and not undertaking "genetic testing and confessionals" on all subjects. We believe that this criticism is unwarranted and stems from a misunderstanding of the purpose of our study. As stated in our paper, the study was intended to be purely descriptive in nature. We stated that the purpose of the study was to "characterize the fracture patterns and clinical information relied upon by clinicians at the time of diagnosis in a sample of infants and children with OI."¹ While we hypothesized that there would be pattern difference would exist, we were careful not to state that our study was designed to demonstrate that. As fracture patterns at the time of diagnosis of OI in children had not been described prior, we felt, and still do, that there is value to begin to identify if a true overlap of fracture pattern does indeed exist. Our data, while limited to a retrospective chart review, did not demonstrate any similarity to fracture patterns often described in infant victims of abuse. We recognize that our statement 'infants with multiple rib fractures are unlikely to be suffering from undiagnosed OI' may seem generous to Dr. Shelmerdine and colleagues. Given that our study is the only one to report fracture patterns in infants at the time of diagnosis of OI in the literature and that no new data has emerged in the 2 years since publication, that conclusion remains unchallenged. As noted by Dr. Shelmerdine and colleagues, we did identify 22% of the subjects did indeed have rib fractures, with 13/15 presenting antenatally or in the immediate newborn period. None of our subjects developed rib fractures in infancy (28 days to 12 months of life). As we noted in our response to Dr. Paterson's letter^{3,4}, birth is a profoundly traumatic experience and it remains unlikely that an infant with a predisposition to fractures would be born without skeletal injury, but then readily fracture as an infant.

Along these lines, Dr. Shelmerdine and colleagues express concern that our study will be used to “to refute an alternative diagnosis in a child with multiple rib injuries.”² While the judicial process can torture published data beyond recognition, we were careful not to imply that OI is not a serious consideration when evaluating a child with seemingly unexplained fracture. OI is a meaningful consideration in all infants and children with seemingly unexplained fractures. The purpose of the study was to begin to explore how similar, at the time of the diagnosis of OI, these conditions really were. The first step, our study, is to describe the fracture pattern at the time of OI diagnosis. In that context, we are unclear what Dr. Shelmerdine and colleagues meant regarding alternative diagnoses in children with multiple rib injuries. If there exist data demonstrating that children at the time of diagnosis with OI can present (outside of the immediate newborn period) with multiple rib fractures, we are unaware of it.

Second, Dr. Shelmerdine and colleagues indicate that OI and physical abuse of a child are not mutually exclusive. We agree. They further interpret our paper as implying that we were stating that we could indicate if a fracture were intentional or accidental based upon radiographic imaging. We regret that we may have given Dr. Shelmerdine and colleagues that impression, but are unclear where in the manuscript that was implied. Again, as a descriptive study of fracture patterns in children at the time of diagnosis of OI, we in no way were exploring causes or mechanism of injury; accidental or inflicted.

Lastly, Dr. Shelmerdine and colleagues also are critical of our paper in not presenting radiographic imaging. We agree and as we noted such in our manuscript. We urged caution in over interpreting our data stating “we likely have undercounted the number of fractures because as a record review we did not review the radiographs individually and some subjects may have not had complete skeletal assessments at the time of diagnosis.”¹

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