Dominant inheritance of primary glenoid dysplasia: report of two cases

Stuart N. Andrews*, Tim D. Bunker*, and Sarah F. Smithson**

*Princess Elizabeth Orthopaedic Centre, Royal Devon & Exeter Hospital, Barrack Road, Exeter EX2 5DW, UK; and **Department of Clinical Genetics, St Michael’s Hospital, Bristol, BS2 8EG, UK

Corresponding address: T. D. Bunker, Elizabeth Orthopaedic Centre, Royal Devon & Exeter Hospital, Barrack Road, Exeter, EX2 5DW, UK.

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Abstract

Primary glenoid dysplasia, although a relatively rare diagnosis, has been well described clinically and published in the orthopaedic literature. However, very few cases of familial occurrence of the disease have been reported. We present two cases of the condition in a father and son. Both presented in childhood with recurrent shoulder pain and radiologically display primary glenoid dysplasia. Our cases would strongly suggest autosomal inheritance may occur in this dominant condition.

Keywords

Primary glenoid dysplasia; Autosomal inheritance.

Case 1

A 25-year-old male, who presented in childhood with recurrent shoulder pain, had a frank dislocation of the left shoulder at the age of 17 while playing rugby. The radiology at this time identified that he had abnormal glenoid fossae and the diagnosis was made. Since the dislocation, inability to use the left shoulder due to severe pain and limited range of movements in all directions forced him to give up his job. The right shoulder had a less severe restricted range of movement and was considerably less symptomatic but also showed radiological evidence of primary glenoid dysplasia (Fig. 1). A bone block procedure was performed on the left shoulder.

Case 2

The patient’s son presented at 3 years of age with recurrent shoulder and knee pain. There were no episodes of dislocation and the range of movement was not restricted at either shoulder joint. Radiology at this point showed that he was also affected by shallow glenoid fossae bilaterally (Fig. 2).

When considering our family of father and son we discovered that the father of case 1 had reportedly suffered from shoulder problems and a restricted range of movement at the shoulders all his life, but passed away without any radiological evidence of dysplasia. The sister of case 1 was symptom free and radiologically had normal shoulders.

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Primary glenoid dysplasia often occurs sporadically but there have been several reports of familial recurrence all consistent with autosomal dominant inheritance\cite{1-5}. In one of these families\cite{5}, a young woman with normal scapulae had an affected son, daughter and brother. The observation that obligate gene carriers can be clinically unaffected suggests that this gene may have variable penetrance within families. The family we describe with an affected son and father (and probably grandfather), provide further evidence that (at least in some cases) this is a single gene disorder with autosomal dominant inheritance. At present the gene is unknown and linkage analysis (to locate the gene) has so far been precluded by the relatively small number of families reported. It is interesting that this gene seems to have a localised effect on the development of the scapula especially the glenoid fossa.

These familial cases emphasise the importance of taking the family history in this condition. If there is already an affected parent and child within a family, the risk of another child (male or female) inheriting the gene is 50%. At present it is unknown what percentage of cases of primary glenoid hypoplasia are genetically determined and, if so, how many are due to errors in the same gene. However, it is likely that some apparently isolated cases without a family history (due to new spontaneous mutations) will also be at risk of having an affected child. Definition of the condition at the molecular genetic level will help to address these questions in the future.
References