

OBJECTIVES

To ascertain the incidence of arthrogryposis multiplex congenital among children in Australia, identify the group(s) of clinicians who care for them, describe the pattern of malformations among these children and possible influence of parental/genetic factors.

INVESTIGATOR CONTACT DETAILS (*Principal Investigator)

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SUMMARY PROTOCOL

Arthrogryposis multiplex congenital (AMC) is used to describe '**conditions with multiple non-progressive congenital joint contractures**'. The incidence is 1-3 per 10,000 liveborn infants and 26-78 new cases of AMC would be expected to occur among Australian livebirths each year. A range of conditions involving multiple joint contractures may be included under a diagnosis of AMC. It is therefore important to ascertain the profile of specific birth defects labelled as AMC for surveillance purposes and to assist with the development of a more precise classification of such cases, if necessary.

The NSW Birth Defects Register (BDR) is a population-based surveillance system which monitors birth defects detected during pregnancy or diagnosed in infants up to one year of age. The BDR is based in the Epidemiology Branch of the NSW Health Department. Its purpose is to provide information to support planning and evaluation of health services and to provide an 'early warning system' which may be used for the detection of previously unrecognised teratogens. Notification of birth defects to the BDR is currently voluntary. For 1990-91, 5 liveborn cases of arthrogryposis were notified to the BDR. This compares with about 8-24 cases which would be expected in NSW for the two years. Thus AMC is under-enumerated by the BDR.

This study aims to determine the true incidence of AMC for Australia and each Australian State/Territory, identify the group(s) of clinicians who care for cases of AMC, compare the ascertainment of AMC in NSW using the APSU and the BDR, and to describe the range of conditions labelled as AMC. The latter will assist in modifying the definition and classification of AMC for surveillance purposes.

CASE DEFINITION

Any child born on or after 1/1/96 with **two or more non-progressive joint contractures present since birth**.

A joint contracture is defined as a condition of fixed high resistance to passive straightening of the joint which is present at birth. This case definition excludes children with unilateral or bilateral talipes where there is no other joint involvement.

REPORTING INSTRUCTIONS

Please report any child with AMC who meets the case definition, who has been seen in the last month and has not previously been reported.

FOLLOW-UP OF POSITIVE RETURNS

A questionnaire requesting further details will be forwarded to the practitioners who report a case.