

# Cervical Vertebral Body Shape and Morphology in Persons with Williams syndrome



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## INTRODUCTION

Williams syndrome (WS) is a rare congenital disorder with distinctive facial features, cardiovascular abnormalities, short stature, mental retardation, and cognitive characteristics. The incidence is estimated to be about 1 in 15-20,000 live births. It is equally prevalent in both sexes and all populations throughout the world. Over 90 per cent of individuals with the clinical diagnosis of WS have a microdeletion on chromosome 7 (del7q11.2).

The size and morphology of the neurocranium and the cranial base differ from normal controls; the anterior and posterior cranial base lengths are shorter, a flattening in the superior aspect of the parietal bone is noted, the posterior part of the occipital bone is more prominent, which is reflected in a larger total length of the neurocranium, and the theca cranium is thicker. (Axelsson S *et al.*, 2005a, 2005b).

## RESULTS

The results show that the vertebral body height vs. depth ratio (H/D ratio) in WS was similar to the values from the reference material. However, for certain age groups the H/D ratio was significantly larger, illustrated by the measurement from C4 on the female group (Figure 3). The sagittal canal diameter vs. vertebral body depth ratio (S/D ratio) in the WS group was over-

all smaller, especially in the male group (Figure 4).

Morphological cervical vertebral anomalies, such as block vertebrae or fusion of C2-C3 (14.5 per cent) (Figure 5), discal narrowing of C2-C3 (8.1 per cent) (Figure 6), and/or spinal stenosis (4.8 per cent), were found in more than 27 per cent of the study group. In the

general pediatric population the occurrence of cervical morphological abnormalities are rare; block vertebrae are found in 4.1 per cent (Uğar and Semb, 2001). The incidence and prevalence data of spinal stenosis is not available but is estimated to be about 5 per cent. The prevalences for block vertebrae, discal narrowing and spinal stenosis in the adult population are not available.

## AIM

The aim of the study was to analyse size and morphology of the cervical vertebrae in persons with WS on lateral cephalograms.

## MATERIAL AND METHODS

The cervical vertebral (C2-C5), body height (H), depth (D) and sagittal canal diameter (S) (Figure 1) were analysed on lateral cephalograms in 62 individuals (25 male and 38 female) with WS, with age range 4.7 to 44.4 years (mean age 14.1 years). The results were compared with normal values of cervical vertebral size for different age groups (Remes VM *et al.*, 2000). The total study sample comprised 105 lateral cephalograms (41 males and 64 females). The distribution of the lateral cephalograms divided into sex and age groups are found in Figure 2.

Figure 1

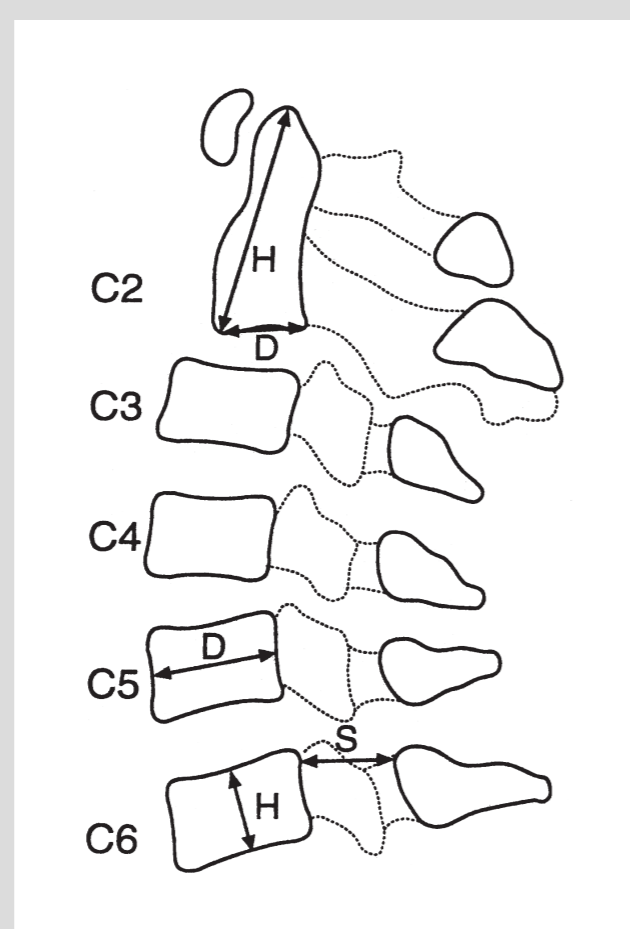
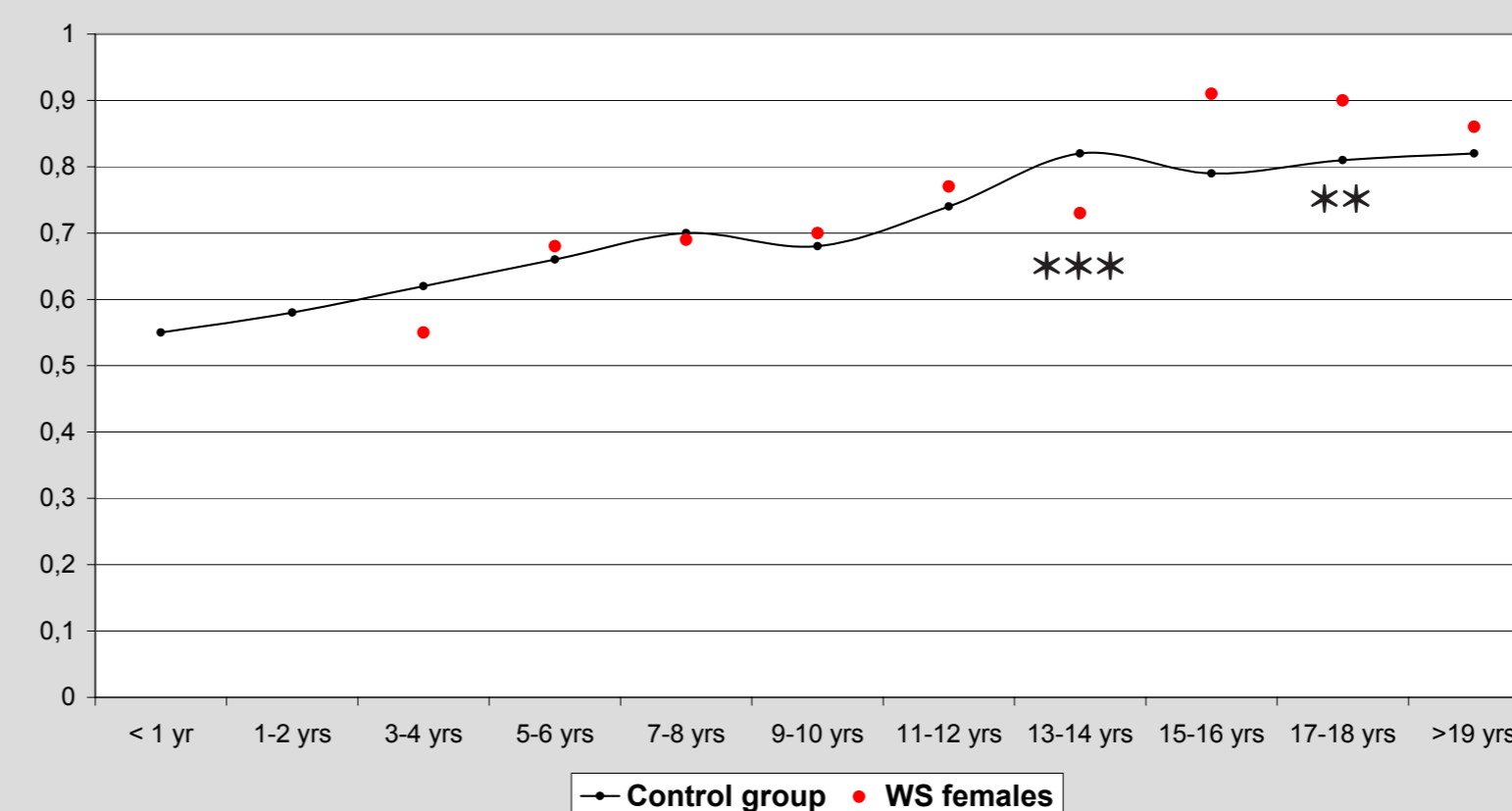


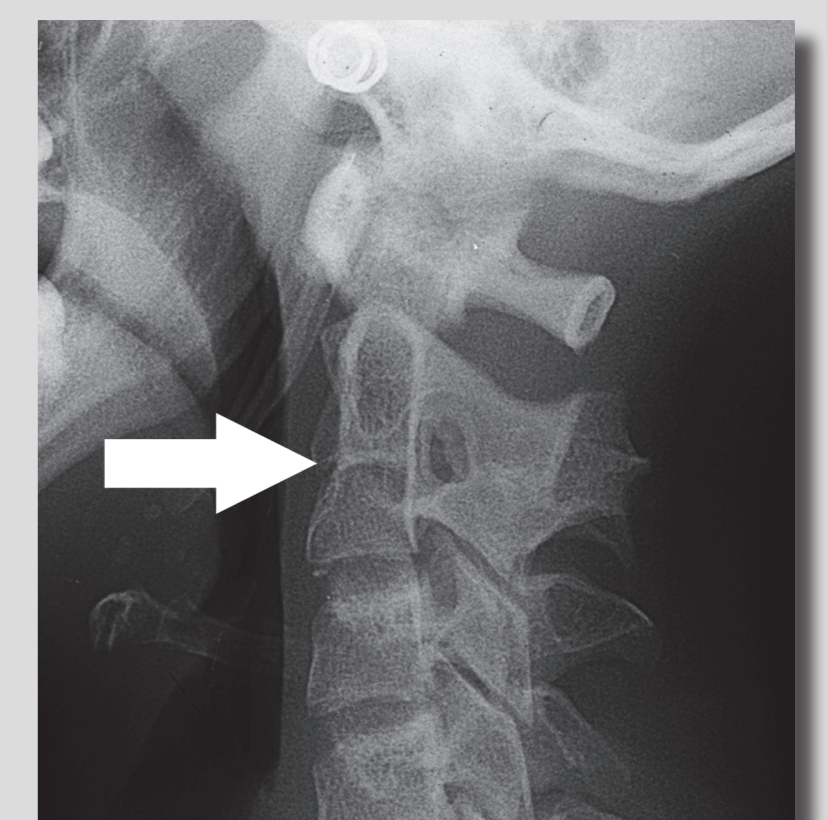
Figure 3

C4 H/D



\*\* significant at  $p < 0.01$  \*\*\* significant at  $p < 0.001$

Figure 5



Block vertebrae C2-C3

Figure 2

Age-distribution

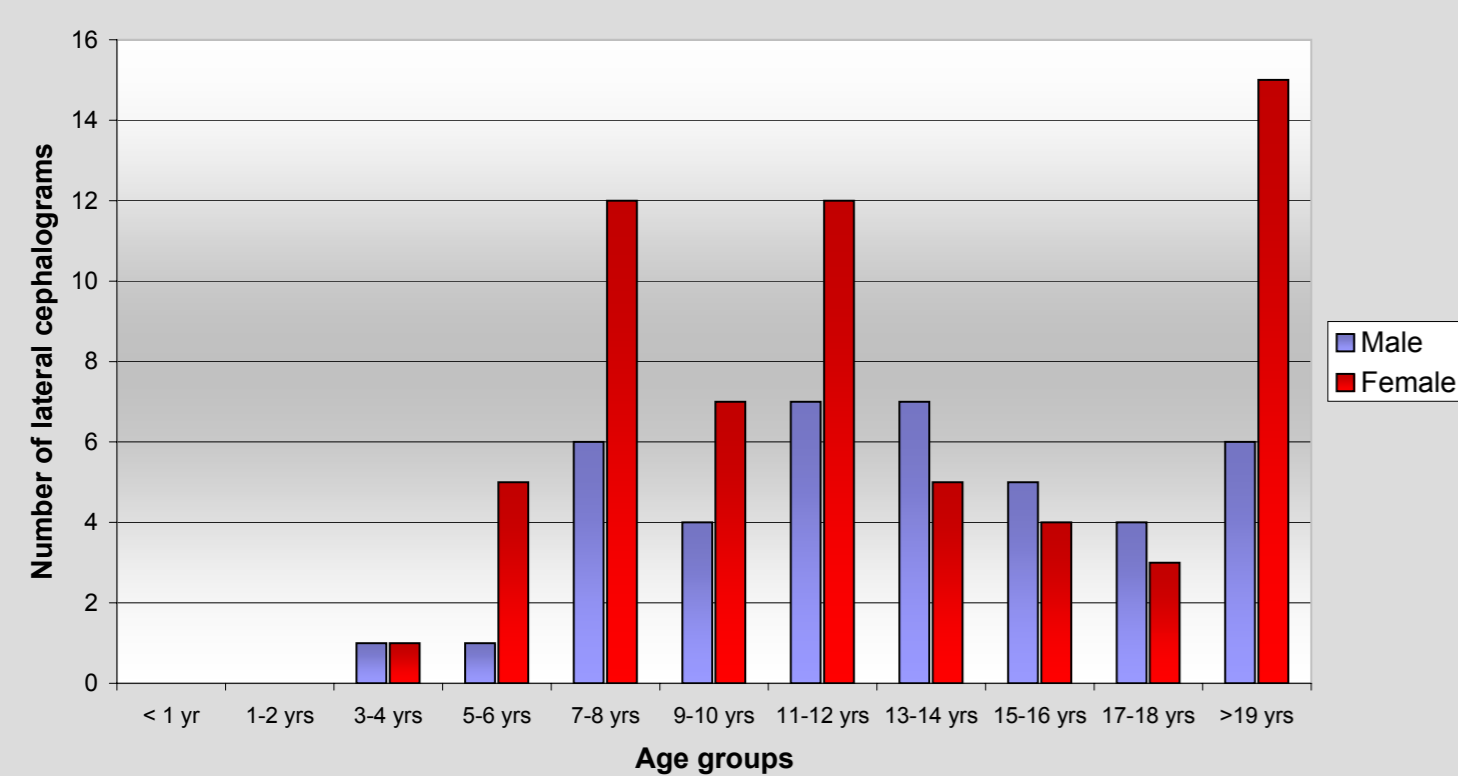
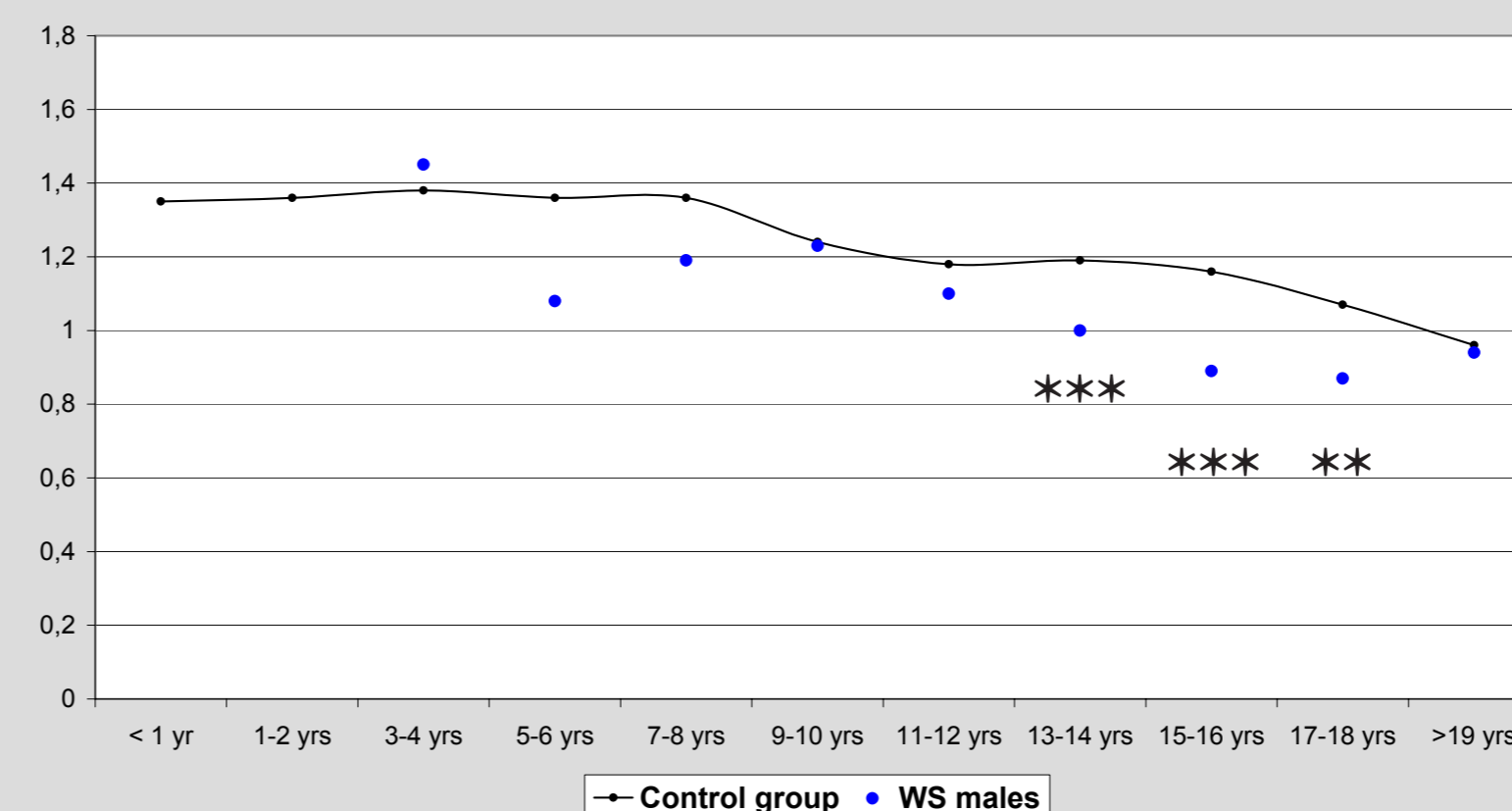


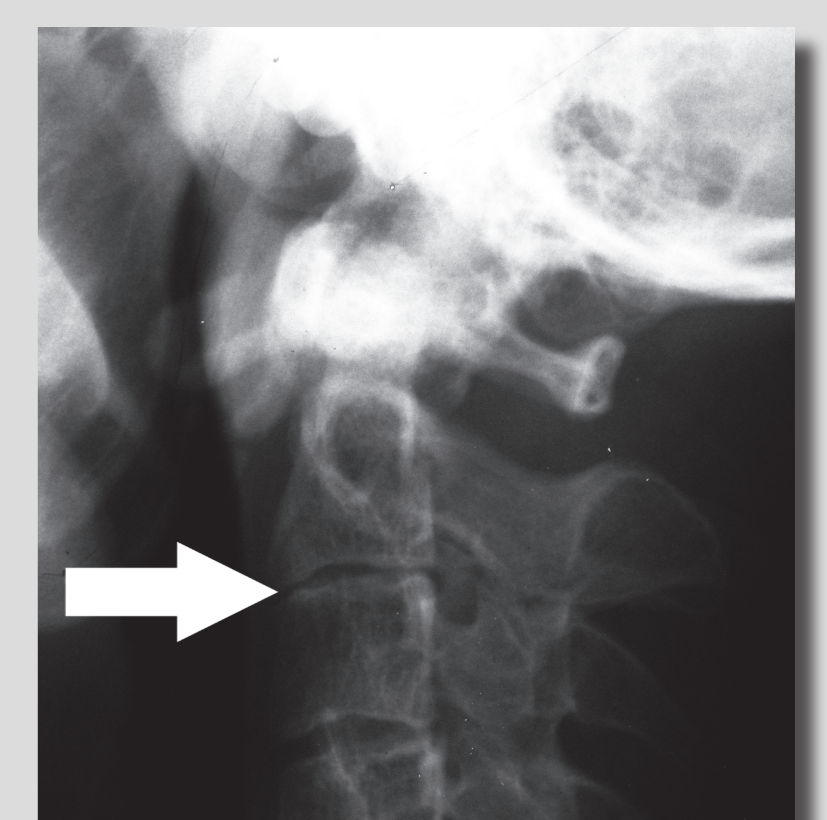
Figure 4

C4 S/D



\*\* significant at  $p < 0.01$  \*\*\* significant at  $p < 0.001$

Figure 6



Discal narrowing of C2-C3

## REFERENCES

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## CONCLUSIONS

The vertebral column is, in its early formation, closely related to the presence, location and later disappearance of the notochord. When the course and/or function of the notochord are abnormal it may be assumed that the skeleton associated with the notochord could be malformed.

Block vertebrae are the results of improper segmentation of the superior and inferior portions of adjacent vertebrae, causing a single

continuous vertebral body composed of two segments. Block vertebrae may cause reduced mobility in extension, flexion and lateral movement of the neck. Discal narrowing and spinal stenosis may give neurological symptoms.

Rapidly progressing scoliosis and kyphosis have been reported in WS. There is, however, only one case report describing cervical vertebrae anomalies in WS in the literature (Ezzeddine *et al.*, 2000).

Variation in size of the cervical vertebrae is common in WS and in some individuals morphological anomalies such as block vertebrae, discal narrowing and spinal stenosis were found. It is premature to predict the clinical implications of these radiographic findings, but the use of advanced imaging techniques and further longitudinal observations may clarify their clinical importance.