Frequencies of neurological malformations in EDS patients

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Methods (cont’d)

foramen magnum. This causes the tonsils to put pressure on the spinal cord resulting in a variety of bone deformities that, when paired with hEDS, can compromise the connection between the skull and spine. To determine the presence of CM-1, a line was drawn from the inner margins of the foramen magnum to the inferior part of the tonsils (see Figure 3); if the length was 5mm or greater, the patient had CM-1. To test for craniocervical instability, a line was drawn down the base of the skull followed by a second line drawn down the back of the body to create the clivo-axial angle. A “normal” clivo-axial angle (Henderson et al., 2010) is approximately 165° when neutral, 155° when flexed, and 175° when extended (see Figure 4). If the clivo-axial angle is less than 135°, the patient has craniocervical instability. Cervical instability was assessed for by measuring the curve in the cervical spine; if there was a reverse, or kyphotic angle, the patient had cervical instability. Originally, the plan was to compare the neurological malformation frequencies discovered in this study to their counterparts in the general population, however after extensive research through various medical databases and literature, it was discovered that there are no known set frequencies of craniocervical or cervical instability in the general population, only of CM-1 (one in every thousand births). The fact that there is so little known about those disorders was a discovery in it of itself. To continue with the project, the results from a study comparing neurological malformations in male and female patients with Down syndrome conducted by El-Khouri et al. (2014) were utilized. Rather than being compared to their frequencies in the general population, the frequencies of cervical and craniocervical instability were compared to their frequencies in Down syndrome patients.

Results

On the basis of two-proportion z-tests, there was convincing evidence that the proportion of CM-1 is significantly higher in hEDS patients than it is in the general public (p-value approx. 0). Cervical instability also appears significantly more frequently in hEDS patients than it does in Down syndrome patients (p-value approx. 0). However, there was no significant difference found between the frequencies of craniocervical instability in hEDS and Down syndrome patients (p-value approx. 0.75).

Conclusion

The prevalence of patients with CM-1 and hEDS is significantly higher than patients in the general public with CM-1. Similarly, the prevalence of patients with cervical instability as well as hEDS is significantly higher than the prevalence of cervical instability in patients with Down syndrome. With further studies, there is a chance that CM-1 and/or cervical instability could be proven to be symptoms of hEDS. Until then, the research conducted in this study means that as soon as a patient is diagnosed with hEDS, a doctor would be within his/her jurisdiction to have the patient tested for CM-1 and/or cervical instability. This would in turn allow patients to be diagnosed before their symptoms become out of hand. Earlier diagnoses would mean preventative measures could be taken to keep the neurological malformations from reaching their maximum irregularities. The patients would ultimately be in far less pain. There was an insignificant prevalence of patients with Down syndrome and hEDS.

References


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