

Scoliosis in association with the 22q11.2 Deletion Syndrome

The 22q11.2 Deletion Syndrome (22q11.2DS) is characterized by a wide variability of clinical features. These features include congenital heart disease (CHD) and scoliosis. A scoliosis is a three dimensional deformity of the spine (a S- or a C-shape), which can develop throughout growth. Little is known regarding how many patients with 22q11.2DS develop a scoliosis. If a scoliosis progresses either brace therapy or scoliosis surgery can be needed.

We performed this study in two specialized 22q11.2DS centers: the Children's Hospital of Philadelphia (CHOP) and the University Medical Center Utrecht (UMCU).

Scoliosis can develop throughout growth and therefore, within the Children's Hospital of Philadelphia, the group of patients older than 16 years of age (317 patients) was analyzed. Forty-eight percent of the patients had a scoliosis. The same was shown for Utrecht, in which 49% (a total of 97 patients) had a scoliosis. The majority of the patients had a small scoliotic curve, which needed no treatment. In total, 8% of the 22q11.2DS patients older than 16 years needed surgery for their scoliosis. In both groups gender was not associated with the occurrence of a scoliosis. Moreover, scoliosis was not associated with the presence of a congenital heart defect. To summarize, nearly half of the 22q11.2DS develop scoliosis and therefore it is an important feature which is definitely associated with the 22q11.2 Deletion Syndrome.

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