Dyggve-Melchior Clausen Disease

Other names that may be used for this disorder are:
- DMC Disease
- DMC Syndrome
- Smith-McCort Dysplasia (variant)

Dyggve-Melchior Clausen Disease is a rare genetic disorder which is characterised by abnormal skeletal development and learning difficulties and developmental delay. The disorder is believed to be caused by a defect in the dymeclin gene which is located on the long arm of chromosome 18. The role of this gene is currently unknown. There is a variant of this disorder known as Smith-McCort Dysplasia which is similar but individuals do not present with mental deficiencies. It is believed that this variant is caused by different defects in the same gene. Approximately 60 cases have been reported in the medical literature.

Treatment for individuals with this disease aims to provide relief for any symptoms and support in the care of the individual. Surgery may be required to correct skeletal abnormalities especially in the spine. Special education may help to enable the child reach his or her full potential. Genetic counselling is recommended for those affected by this disorder and their families.

This information is fully sourced and referenced, for more detailed information and references please contact CLIMB by email, letter or telephone.

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