

Anesthesia recommendations for patients suffering from **Phocomelia**

Disease name: Phocomelia

ICD 10: Q71.1 (phocomelia upper limb), Q72.1 (phocomelia lower limb), Q73.1 (phocomelia unspecified limb)

Synonyms and associated diseases: Roberts SC-Phocomelia syndrome, Roberts Tetrachomelia syndrome, SC Phocomelia syndrome, Pseudo-thalidomide syndrome, Tetrachomelia syndrome, DK Phocomelia, Fuhrman syndrome, Holt-Oram syndrome, Steinfeld syndrome

Phocomelia is a rare birth defect characterised by severe limb deformities. Bones of the upper limbs are shortened or absent and fingers may be fused. In extreme cases, both upper and lower limb bones are absent so that the hands and feet are attached to the body directly constituting what is known as tetrachomelia.

Phocomelia can either be genetically inherited or drug-induced. In its genetic form it is transmitted as an autosomal recessive trait which is linked to several chromosomal mutations. Spontaneous gene mutations have also been recognised.

Drug-induced cases of phocomelia syndrome can be mostly associated with maternal ingestion of the drug thalidomide by expectant females (sometimes referred to as thalidomide syndrome). Thalidomide was developed in Germany in 1954 and gained widespread use as treatment for morning sickness in early pregnancy. It was also used as a sedative and cough suppressant. It has been recently used in the management of multiple myeloma, erythema nodosum leprosum and is also being investigated for the treatment of a variety of other conditions including inflammatory and immunological diseases and some cancers. It was recognised as a cause for severe congenital deformities and was withdrawn from the market worldwide between 1961 and 1962.

Medicine in progress



Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong



Find more information on the disease, its centres of reference and patient organisations on Orphanet: www.orpha.net

Typical surgery

There is no specific surgery undertaken for phocomelia. Corrective surgery, however, maybe required for associated anomalies, for example: craniofacial or limb deformities or scoliosis repair in the more extreme Robert syndrome or SC phocomelia.

Type of anaesthesia

There are currently no recommendations in literature regarding the optimum management of these patients. A variety of corrective surgical procedures may be undertaken for congenital malformations associated with phocomelia.

In adulthood, however, undergoing surgery for incidental conditions (for example acute abdominal conditions), both general and regional anaesthesia can be safely administered. The challenges facing the anaesthetist are numerous and include:

1. Difficulty in monitoring blood pressure: It maybe impossible to measure the blood pressure non-invasively due to the absence of limbs or if they are attached to the trunk via very short appendiges. Invasive blood pressure monitoring may itself be very challenging too. The choice of arteries available maybe limited to the femoral and axillary arteries which maybe aberrant in course and caliber causing major difficulties in accessing them. Alternative approaches for assessment of cardiovascular status to enable fluid management may need to be resorted to, including non-invasive cardiac output monitoring (3).
2. Difficult venous access: This can present a huge challenge as only central veins maybe accessible. Early involvement of the anaesthetic team should be considered to secure central venous access in these patients. In these cases ultrasound can be very helpful.

Necessary additional diagnostic procedures (preoperative)

Depending on the associated diseases or congenital malformations, specific investigations maybe required, for example, detailed cardiac function assessment would be required in the presence of cardiac defects, pulmonary function test in the presence of severe scoliosis and special preparations maybe needed for airway abnormalities.

Particular preparation for airway management

In cases where airway abnormalities are evident or when difficult airway is expected, advanced planning, specialist instruments and appropriately trained medical staff must be available. Be aware of that some forms of phocomelia have been associated with micrognathia.

Particular preparation for transfusion or administration of blood products

The need for transfusion will be dictated by the surgical procedure undertaken. Appropriate preparation for difficult venous access must be addressed.

Thrombocytopenia can be associated with phocomelia

Particular preparation for anticoagulation

Anticoagulation requirements will depend on the surgical procedure and the length of immobility in the postoperative period. Some patients may display coagulation defects which must be managed on an individual case basis.

Particular precautions for positioning, transport or mobilisation

Phocomelia patients have mobility restrictions and require assistance in transport to theatre. Special handling and positioning may be required.

Probable interaction between anaesthetic agents and patient's long term medication

Not reported.

Anaesthesiologic procedure

Both general and regional anesthesia is appropriate. Difficulties in monitoring and vascular access should be addressed as pointed out above.

Particular or additional monitoring

Invasive or non-invasive cardiovascular status monitoring could be required to assist fluid management in the perioperative period.

Possible complications

As phocomelia can be associated with a variety of other pathologies, the complications encountered will be dependent on the degree of other organs' in the disease process.

Postoperative care

Depending on the surgical intervention undertaken, and the associated defects in patients with phocomelia, these may require nursing in a high dependency environment postoperatively to allow for close monitoring of physiological parameters.

Information about emergency-like situations / Differential diagnostics

caused by the illness to give a tool to distinguish between a side effect of the anaesthetic procedure and a manifestation of the diseases, e.g.:

As phocomelia in itself does not have systemic manifestations, there is nothing specific in this category.

Ambulatory anaesthesia

Since phocomelia is frequently associated with other congenital anomalies, it is more common to resort to general anaesthesia. There is, however, no contraindication to the use of ambulatory anaesthesia if it is safe and practical to do so.

Obstetrical anaesthesia

There are very few reports of anaesthetising females with phocomelia for delivery but successful regional anaesthetic techniques have been used without incident. Obvious consideration must be made in patients with congenital spinal deformities in which central neuraxial block may be challenging.

Literature and internet-links

1. National Organization for Rare Disorders. <http://www.rarediseases.org>
2. History of Thalidomide. <http://www.thalidomidesociety.co.uk>
3. K Mukhtar, S Jayaseelan, E Allsop. Phocomelia: a lesson from the past. *Eur J Anaesthesiol* 2012;29(7):353-4
4. S Blacker, T Angelo. Anesthetic management of a parturient with Thalidomide phocomelia. *Open Journal of Anesthesiology* 2012, 2: 59-61
5. Bermejo-Sanchez E, Cuevas L, Amar E, Bianca S, Bianchi F, Botto LD, Canfield MA,
6. Castilla EE, Clementi M, Cocchi G, Landau D, Leoncini E, Li Z, Lowry RB, Mastroiacovo P, Mutchinick OM, Rissmann A, Ritvanen A, Scarano G, Siffel C, Szabova E, Marti'nez-Fri'as M-L. 2011. Phocomelia: A worldwide descriptive epidemiologic study in a large series of cases from the International Clearinghouse for Birth Defects Surveillance and Research, and overview of the literature. *Am J Med Genet Part C Semin Med Genet* 157:305–320.
7. Louise Harewooda, Jean W. Keelingb, Judith A. Fantesa, John M. Opitzc and David R. Fitz Patricka . Crommelin-type' symmetrical tetramelic reduction deformity: a new case and breakpoint mapping of a reported case with de-novo t(2;12)(p25.1;q23.3). *Clinical Dysmorphology* 2010, 19:5–13
8. Lubinsky MS, Kahler SG, Speer IE, Hoyme HE, Kirillova IA, Lurie IW. Von Voss-Cherstvoy syndrome: a variable perinatally lethal syndrome of multiple congenital anomalies. *Am J Med Gene.* 1994 Sep 1;52(3):272-8
9. Kristin Becker, Karol Howard and Helen Hughes. DK–phocomelia syndrome with thrombocytopenia, encephalocele, and choanal atresia in an adult male with moderate learning difficulties. *Clinical Dysmorphology* 2011, 20:152–155

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