orphanaesthesia

Anaesthesia recommendations for

Phocomelia

Disease name: Phocomelia

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

Synonyms: Roberts SC-Phocomelia syndrome, Roberts Tetraphocomelia syndrome, SC Phocomelia syndrome, Pseudo-thalidomide syndrome, Tetraphocomelia syndrome, DK Phocomelia, Fuhrman syndrome, Holt-Oram syndrome, Steinfeld syndrome.

Disease summary: 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 tetraphocomelia. 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 considered for the treatment of a variety of other conditions including inflammatory and immunological diseases and some cancers. Despite its known teratogenic side effects, thalidomide is still currently prescribed because of its immunomodulatory and anti-inflammatory properties. This has led to the emergence of new cases of thalidomide embryopathy, particularly in areas with endemic leprosy.

Medicine is in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnosis 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, may be required for associated anomalies, for example: craniofacial or limb deformities or scoliosis repair in the more extreme Robert syndrome or SC phocomelia. It should also be noted that, due to the current advances in care, there are increasing numbers of adults affected by phocomelia that present with medical conditions that require emergency interventions.

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, especially when dealing with patients who present phocomelia as part of a syndrome, are numerous and include:

1. Difficulty in monitoring blood pressure: It may be 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 appendages. 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 may be accessible. Early involvement of the anaesthetic team should be considered to secure central venous access (or a PICC line) in these patients, preferably on the day prior to the surgical procedure and in a high dependency environment. In these cases, ultrasound can be very helpful.

3. Difficult intubation: Limited neck movement due to cervical spine abnormalities may be encountered

4. Difficult regional techniques: Spinal deformities, including anterior fusion of thoracic and lumbar vertebrae, spina bifida and scoliosis, may render spinal or epidural anaesthesia more challenging.

Necessary additional pre-operative testing (beside standard care)

Depending on the associated diseases or congenital malformations, specific investigations may be 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 may be needed for airway abnormalities. It is crucial that early and detailed pre-operative assessment is undertaken for patients undergoing elective surgery to

identify potential issues and ensure that adequate facilities are available for peri-operative care, including intensive post-operative rehabilitation.

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 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, but it should be noted that, due to a reduced skeletal muscle mass which acts as a vascular reservoir, the need for blood transfusions may be increased. Appropriate preparation for difficult venous access must be addressed and central venous access should be considered.

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 post-operative period. Some patients may display coagulation defects which must be managed on an individual case basis.

Particular precautions for positioning, transportation and mobilisation

Phocomelia patients have mobility restrictions and require assistance in transport to theatre. Special handling and positioning may be required due to the prevalence of joint dysplasia.

Interactions of chronic disease and anaesthesia medications

Not reported.

Anaesthetic procedure

Both general and regional anaesthesia are 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 involvement of other organs in the disease process.

Post-operative care

Depending on the surgical intervention undertaken and the associated defects in patients with phocomelia, these may require nursing in a high dependency environment post-operatively to allow for close monitoring of physiological parameters. Many patients may need intensive physiotherapy post-operatively and may require non-standard, special equipment that will need to be ordered and be available prior to the surgical procedure.

Disease-related acute problems and effect on anaesthesia and recovery

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

Although there are very few reports of anaesthetising females with phocomelia for delivery, 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.

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