orphanaesthesia

Anaesthesia recommendations for

Fabry disease

Disease name: Fabry disease

ICD 10: E75.2

Synonyms: Morbus Fabry, Anderson Fabry disease, Fabry syndrome, Angiosarkoma corporis diffusum, α-galactosidase A deficiency

Disease summary: Fabry disease (FD) is a rare lysosomal storage disease of X-linked recessive inheritance, first described in Germany and the United Kingdom in 1898 [1-3]. Due to mutations in the GLA gene, located on the X chromosome (Xq22.1), patients show a partial or complete deficiency of ceramidtrihexosidase, also referred to as α-galactosidase A (α-Gal A) [3, 4]. The biochemical aetiology of this condition was discovered several years later [5, 6]. As a result of GLA mutations, sphingolipid accumulation occurs in various tissues, with globotriaosylceramide (Gb3) predominantly depositing in the skin, eyes, heart, kidneys, brain, and both the vascular and nervous systems [3]. Accordingly, FD is a multisystem disease.

FD can be categorized into a severe, classical phenotype and a generally milder, non-classical phenotype. In the severe form, enzyme activity is typically absent or negligible [4]. The non-classical type, also referred to as late-onset or atypical FD, exhibits greater variability in disease severity and progression, with disease manifestations often limited to a single organ, most commonly the kidneys or heart [4]. Males tend to develop greater disease severity than females [3]. Skewed X inactivation might be responsible for the variability of the phenotype in women [4, 7].

The incidence of Fabry's disease has been estimated at 1/40 000 to 1/117 000 worldwide up to much higher incidence with about 1/3100 to 1/1000 in high-risk populations and even 1:875 in male and 1:399 female live births in Taiwan. It seems to differ between various countries [8-14].

In a cohort of 98 male patients, the mean age of diagnosis was 21.9 years [15]. The mean median cumulative survival seems to be 50 ± 8 years for males and up to 72 years for females [15-17]. The disease may present at any age and generally is progressive [18].

Diagnosis can be challenging, as patients often present with nonspecific symptoms such as headaches, limb or abdominal pain, and diarrhoea. In most cases, a definitive diagnosis by genetic testing is established only after the occurence of severe complications, including stroke, heart and kidney failure [19]. Blood tests can be used for screening and monitoring (e.g. measurement of α-galactosidase A enzyme activity, plasma lyso-Gb3 levels as a biomarker for disease activity) [20].

Typical facial features include periorbital fullness, prominent lobules of the ears, bushy eye-brows, recessed forehead, pronounced or prominent nasal angle, generous nose or bulbous nasal tip, prominent supraorbital ridges, shallow midface, full lips, prominent nasal bridge, broad alar base, coarse features, posteriorly rotated ears and prognathism [19]. Other features

include short fingers, prominent superficial vessels of hands, 5th digit brachydactyly or clinodactyly [19].

Cardiac abnormalities are a common manidestation of FD, affecting up to 60% of male patients with the classical form [21]. The predominant finding is concentric left ventricular hypertrophy (LVH). Left ventricular mass index seems to be inversely correlated with α-Gal activity [22]. Moreover, Fabry patients often present with left sided valvular dysfunction and conduction disturbances (bradycardia, atrioventricular block, various forms of ventricular and supraventricular arrhythmias, in particular atrial fibrillation) [21, 23, 24]. Patients with FD may therefore be fitted with pacemakers or ICDs [20]. Although less common, right ventricular involvement can occur, leading to both systolic and diastolic dysfunction [25, 26]. The main cause of death in FD is cardiovascular events (75%), with SCD being the most common cause of death (62%) [20].

End-stage renal disease and cerebrovascular events are relatively common in FD, primarily due to glycolipid deposition in the glomeruli. Chronic kidney disease is characterized by glomerulosclerosis, tubular atrophy, and interstitial fibrosis, ultimately leading to proteinuria and progressive renal insufficiency [27]. In the central nervous system, patient exhibit an increased incidence of stroke [21]. Mild cognitive abnormalities are only rarely described [28].

Pulmonary manifestations include obstructive airflow limitation [29, 30].

Impaired autonomic and endocrine function is common in FD. Gastrointestinal manifestations, including abdominal pain, gastrointestinal dysmotility and episodic diarrhoea, are frequently observed. Dysautonomia due to autonomic neuropathy can furthermore lead to urinary incontinence, and orthostatic hypotension. Besides, patients often report headaches and fever of unknown origin [20, 31].

Reduced saliva and tear production, as well as hypohydrosis, may also occur and contribute to decreased exercise tolerance [18, 32]. Some of these symptoms as well as fatigue, dry skin or nonspecific gastrointestinal complaints overlap with those of hypothyroidism, a common co-finding in patients with FD [33, 34].

Peripheral nervous system involvement in FD often leads to neuropathic pain or painful sensations in the extremities (acroparesthesia), while arthralgia and myalgia may further contribute to a reduced quality of life. These symptoms can be triggered by changes in environmental or body temperature, physical exertion, or emotional stress [20, 31].

Other manifestations of FD include corneal opacities (cornea verticillata), angiokeratoma, tinnitus or hearing loss [35, 36].

In recent years, enzyme replacement therapy (ERT) has become available as a treatment option and may help slow disease progression [3]. The Mainz Severity Score Index (MSSI) was developed as a tool to assess disease severity and monitor the clinical course in response to ERT [37]. Migalastat, a pharmacological chaperone, is an additional oral treatment option for certain gene variants of the disease [38].

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

Emergency information

A	**AIRWAY / ANAESTHETIC TECHNIQUE**	Prepare for difficult airway (dysmorphic features of head, face and neck) – pulmonary manifestations include obstructive airflow obstruction – no general (dis)advantage for GA (TIVA / balanced) or GA – in patients with severe cardiac, renal or other organ dysfunctions, GA techniques might be the optimal approach whenever applicable
B	**BLOOD PRODUCTS (COAGULATION)**	No specific recommendations
C	**CIRCULATION**	Cardiac abnormalities are frequent with predominantly left-sided valvular dysfunction, hypertrophy and conduction disturbances – increased risk for cerebrovascular events – consider IBP and (non-)invasive haemodynamic monitoring
D	**DRUGS**	No risk for MH – consider drug dose adaption in case of renal impairment – continue enzyme replacement therapy – endocarditis prophylaxis (if indicated)
E	**EQUIPMENT**	Perioperative availability of dialysis may be necessary – avoid extreme positioning due to haemodynamic impairment and chronic pain – avoid trigger for (neuropathic) pain exacerbation (i.e. emotional stress, changes in body temperature)

Typical surgery

Dermatology: typical vascular skin lesions termed angiokeratoma [39].

Ophthalmology: corneal opacities (cornea verticillata) and other ophthalmological changes [40].

Cardiac surgery: valve repair or replacement, implantation of a pacemaker or internal cardiac defibrillator, cardiac assist devices, resynchronisation therapy and heart transplantation [18, 21, 41].

Neurology and Neurosurgery: cerebrovascular strokes in small or large vessels [42, 43].

Surgery: arterio-venous fistula for haemodialysis, cadaveric renal transplantation, partial nephrectomy [44, 45].

Traumatology: fractures or injuries due to seizures [21].

Type of anaesthesia

General anaesthesia (GA) and regional anaesthesia (RA) techniques may present specific challenges in patients with FD.

Ideally, these patients should be assessed and managed by a senior anaesthetist. Due to the multisystem involvement of FD, a thorough preoperative assessment is essential, with careful assessment of potential organ dysfunction [46].

GA has been described in only a few patients. The use of fentanyl, propofol, rocuronium, atracurium and cisatracurium for induction has been reported as uneventful [47-49]. However, one case describes Fabry patient with transient bronchospasm following GA induction with fentanyl, propofol, and atracurium, though anaphylaxis was ruled out [50]. In case of preterminal or terminal renal insufficiency, a relevant prolongation of elimination times should be anticipated for certain drugs undergoing renal clearance.

For maintenance sevoflurane, desflurane, propofol, as well as remifentanil, sufentanil and fentanyl (bolus administration) as well as the continuous administration of atracurium have been used without reported complications [46, 49, 50].

Postoperative pain management can be difficult due to the episodic or chronical pain often present in Fabry patients. For analgesia, morphine, metamizole, paracetamol as well as lidocaine have been administered without adverse effects [46, 51].

To minimize postoperative nausea and vomiting dexamethasone and ondansetron have been used reported complications [46].

Uneventful administration of neostigmine and glycopyrrolate have been reported to reverse neuromuscular block [46].

To date, no reports exist on the use of peripheral nerve blocks in patients with FD, with only a single case describing an uncomplicated spinal anesthesia for cesarean section [52]. However, the use of RA techniques might be favourable in patients with relevant organ disorders when applicable. To minimize the risk of inadvertent puncture, careful consideration should be given to the location of angiokeratomas and hemangiomas during pre-procedural planning, including physical examination and imaging if necessary. Ultrasound-guided neuraxial or peripheral RA may serve as a viable alternative. Pre-existing disease-specific neurological conditions, such as hypoesthesia, paresthesia, or neuropathic pain, should be thoroughly documented and monitored. Following neuraxial procedures, the differential diagnosis of post-dural puncture headache in FD patients should always include acute vascular events such as stroke. Additionally, spontaneous spinal epidural hematoma has been reported in FD and should be considered as a differential diagnosis in cases of paralysis or other neurological deficits [53].

Necessary additional pre-operative testing (beside standard care)

There is no general recommendation or protocol for an ideal preoperative assessment. Given the various manifestations and variability of symptoms, preoperative assessment must be tailored to identify the specific pattern of symptoms of each individual patient [46]. Particular attention should be given to detect organ dysfunction, with a focus on pulmonary, cardic, cerebral, and renal involvement [51].

Patients with existing chronic pain require careful consideration of the perioperative pain treatment plan [46].

Preoperative evaluation of functional capacity status using an objective tool is likely to be helpful in identifying significant cardiorespiratory disease (e.g. cardiopulmonary exercise testing, 6 Minute Walk Test) [29].

With regard to frequent cardiac involvement in FD, a 12-lead ECG as well as transthoracic echocardiography may be useful to identify valvular disease and assess global ventricular function [51]. Further diagnostic options are transesophageal echocardiography or cardiac MRI [20]. Some authors recommend a high index of suspicion of occult disease and suggest the use of non-invasive cardiac stress tests in patients over 30 years of age and relevant symptoms [50].

Laboratory analysis is recommended to identify patients with impaired kidney function [51].

Particular preparation for airway management

Airway examination should be performed carefully, with particular attention to patient‘s anatomic and dysmorphic features, especially those affecting the head and neck, to identify potential airway difficulties [51]. Difficult airway management should be anticipated and strategies for airway management should be carefully planned in advance. Prognathism or physical features concerning midface, lips and nose may impair optimal mask ventilation. Pulmonary impairment ould further complicate ventilation. Laryngoscopy and intubation may be challenging [51].

Overall, airway evaluation and preparation in FD should follow common practice standards for airway management.

Particular preparation for transfusion or administration of blood products

No specific recommendations are given. No typical bleeding disorders were reported for Fabry patients.

Particular preparation for anticoagulation

There are no specific suggestions for Fabry disease. Postoperative anticoagulation should be considered based on the patient’s history of cardiac and valvular surgery, arrhythmias, stroke, or other cardiovascular events, in accordance with current recommendations.

Particular precautions for positioning, transportation and mobilisation

Extreme positioning during surgery, such as (reverse) Trendelenburg positioning, may lead to haemodynamic impairment in patients with severe cardiac involvement. Due to chronic pain disorders in some patients, positioning and mobilisation must be tailored on an individual basis.

Interactions of chronic disease and anaesthesia medications

Not reported. Especially enzyme replacement therapy does not interfere with any of the reported drugs used [46].

Anaesthetic procedure

Preoperative Evaluation: see details above.

Premedication: might be performed weighing the benefits and risks in individual patients. Enzyme replacement therapy should be continued following regular prescription when undergoing GA.

Endocarditis prophylaxis: should be administered in patients with an established indication, particularly those with a history of cardiac valve surgery, in accordance the current international guidelines and in consultation with the responsible cardiologist [54].

Patient positioning & monitoring: act with caution due to haemodynamic impairment in case of severe cardiac involvement and long-lasting chronic pain anamnesis.

Vessel cannulation: might be difficult due to vascular impairment or large-area haemangioma [51]. Consider ultrasound-guided vessel cannulation.

Anaesthesia: induction of anaesthesia should be tailored to patient-specific risk factors, with particular consideration of cardiopulmonary involvement. With regard to physical features and pulmonary impairment, difficult airway management should be anticipated, particularly in relation to bag-mask ventilation, laryngoscopy, and intubation. Using established drugs (see details above) for both induction and maintenance has been reported as uneventful. Total intravenous (TIVA) or balanced anaesthesia with volatile anaesthetics appear to be safe. Drug dosage should be adjusted according to renal function.

Particular or additional monitoring

Cardiopulmonary evaluation may include invasive blood pressure measurement and non-invasive cardiac output monitoring to guide intraoperative fluid and blood pressure management. The extent of monitoringshould be tailored to patient's clinical status and the specific surgical procedure following standard perioperative care principles [51]. BIS monitoring can assist in evaluating and adjusting for the maintenance of anaesthesia [49]. Affected patients often have metabolic derangements, with signs of hyperkalaemia being particularly prominent, requiring careful perioperative monitoring and management [49].

Possible complications

Complications in airway management (bronchospasm, laryngoscopy, ventilation) are reported [50, 51]. Postoperative pain management may be challenging. Haemodynamic instability due to underlying cardiovascular impairment is reported.

Post-operative care

Postoperative care should be individualised based on the patient's disease severity and type of surgery. While routine admissionto an intermediate or intensive care unit is not mandatory, it may be warranted in cases of severe organ dysfunctions or when postoperative dialysis is necessary.

Disease-related acute problems and effect on anaesthesia and recovery

Desaturation and hypoxia: allergic genesis, pulmonary embolism, endotracheal disconnecttion or other technical problems.

Haemodynamic deviation: anaphylactic genesis, myocardial infarction, bleeding complications.

Ambulatory anaesthesia

Ambulatory anaesthesia is feasible and can be performed in institutions with adequate resources and expertise. Depending on pre-existing cardiac, respiratory and renal dysfunction and the type of surgery, the decision should be made on a case-by-case basis. A prolonged stay in the post anaesthesia care unit (PACU) may be necessary due to prolongated drug effects. Currently, there are no general recommendations regarding outpatient procedures due to a lack of reports in the literature.

Obstetrical anaesthesia

Patients with FD are fertile. To date, only one report has documented the use of spinal anesthesia in a first-time gravida with FD, describing an uneventful procedure without complications [52]. However, due to a lack of reports on further spinal or epidural anaesthesia in Fabry patients, establishing definitive recommendations remains challenging. Haemangioma can occur over the medial spine and should be considered [55]. There is one case report of a spontaneous spinal epidural haematoma in a non-pregnant woman with FD [53]. While cerebrovascular events are common in FD, no cases of vertebral dissection or spinal cord infarction have been documented in the literature to date [47]. However, in patients without relevant bleeding anamnesis and with normal coagulation parameters, RA may be considered. However, depending on their location, accidental puncture of angiokeratomas and hemangiomas, which are typical of FD, may affect the spread of local anaesthetics near the spinal cord and, in the worst case, have fatal consequences [53, 56]. Few reports involving small cohorts of pregnant women with FD who continued or reinitiated ERT during pregnancy have not identified no adverse effects in either mothers or infants [52, 57]. Migalastat therapy is not recommended during pregnancy [58]. Reported pregnancy-related complications requiring emergency cesarean section include eclampsia with proteinuria, hypertensive crisis and seizures, pathological cardiotocographic findings, and premature birth [55, 57].

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