orphanaesthesia

Anaesthesia recommendations for

Lujan-Fryns syndrome

Disease name: Lujan-Fryns syndrome

ICD 10: Q87.8

Synonyms: Lujan syndrome, X-linked mental retardation with marfanoid habitus, XLMR with marfanoid habitus

Disease summary: Lujan-Fryns syndrome (X-linked mental retardation with marfanoid habitus syndrome) is characterised by a marfanoid habitus becoming evident after puberty, typical craniofacial dysmorphism and behavioural problems. Recently, mutations in the MED12 gene and in the UPF3B gene have already been described as the cause of Lujan-Fryns syndrome. Lujan-Fryns syndrome was first reported by Lujan in 1984 and Fryns offered other features of this syndrome in 1987. Craniofacial features in Lujan-Fryns syndrome include prominent forehead, long narrow face, small mandible, maxillary hypoplasia, long nose with high and narrow nasal bridge, short and deep philtra, thin upper lip, highly arched palate, receding chin, and low-set retroverted normal shaped ears. Complete or partial agenesis of corpus callosum, ascending aorta aneurysm and ventricular septal defects are other manifestations of Lujan-Fryns syndrome. The prevalence in the general population is not known but affects predominantly males. There is no available treatment for Lujan-Fryns syndrome, patients need special education and psychological support. Diaphgram hernia and pulmonary hypoplasia are the most common anomalies in this syndrome. Because of these anomalies. limited pulmonary reserves and marked facial dysmorphism make it difficult to have a secure airway. Furthermore, the presence of cardiovascular malformations can lead to significant problems during anaesthesia practice.

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

	AIRWAY / ANAESTHETIC TECHNIQUE	high risk for a difficult airway due to craniofacial dysmorphies (therefore, GA under ambulatory conditions or outside a major anaesthesiological department is not recommended!) – anticipate limited pulmonary reserves (i.e., diaphragm hernia, pulmonary hypoplasia) – no general (dis)advantage for GA (TIVA / balanced) or RA – RA techniques might be the optimal approach whenever applicable due to high risk of difficult airway
B	**BLOOD PRODUCTS (COAGULATION)**	no specific recommendations
C	**CIRCULATION**	consider echocardiography to detect frequent cardiovascular anomalies (i.e., aortic aneurysm, ventricular septal defect)
D	**DRUGS**	no risk for MH
E	**EQUIPMENT**	consider presence of a (personal) caregiver in PACU / IMC / ICU due to frequent behavioural problems and mental retardation

Typical surgery

There is no typical surgery, but cleft palate surgery, vertebrae surgeries, cardiovascular surgeries can be possible due to accompanying anomalies.

Type of anaesthesia

There is no definite recommendation for either general or regional anaesthesia. However, considering the presence of craniofacial anomalies, which are specific features of this syndrome, the risk of difficult airway during general anaesthesia can make regional anaesthesia preferable.

Necessary additional pre-operative testing (beside standard care)

Most of clinical manifestations can be determined during physical examination, but for the detection of cardiovascular anomalies such as ventricular septal defect and aortic aneurysm, echocardiogram should be part of the routine evaluation.

Particular preparation for airway management

Due to the presence of craniofacial anomalies, these patients are at high risk for a difficult airway.

Particular preparation for transfusion or administration of blood products

Not reported.

Particular preparation for anticoagulation

Not reported.

Particular precautions for positioning, transportation and mobilisation

Not reported.

Interactions of chronic disease and anaesthesia medications

Not reported.

Anaesthetic procedure

In these patients, no problems were encountered after using sevoflurane or propofol. However, adequate information about the use of other anaesthetic agents is not available.

Particular attention should be paid to the anaesthesia practice; it is the likely difficult airway that can be encountered in these patients. All necessary preparations must be made for the difficult airway.

Particular or additional monitoring

Standard monitoring of vital signs should be performed in all types of anaesthesia including sedation.

Possible complications

Complications due to difficult airway scenarios are possible. Prepare for a difficult airway scenario to avoid a "Cannot intubate, cannot ventilate" situation.

Post-operative care

The degree of postoperative monitoring is depending on the surgical procedure and on the preoperative condition of the patient.

Disease-related acute problems and effect on anaesthesia and recovery

As a last resort in a “Cannot intubate, cannot ventilate” situation, an emergency airway-puncture set must be at hand.

Ambulatory anaesthesia

Because of the possible difficult airway in patients with Lujan-Fryns syndrome, and because of the needed additional devices to be prepared, general anaesthesia under ambulatory conditions or outside a major anaesthesiological department cannot be recommended.

Obstetrical anaesthesia

There are no obstetric case reports of patients with Lujan-Fryns syndrome.

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Please note that this recommendation has been reviewed not by an anaesthesiologist and a disease expert but by two anaesthesiologists instead.