

Cystinosis Consultation Checklist

What is this checklist for

This cystinosis-specific checklist is designed to support you in your regular consultations with your adolescent or adult patients. It has the following purposes:

- To help you ensure that all aspects of the multi-organ effects of cystinosis are monitored so that nothing is overlooked as a potential area of concern
- To help you decide whether the patient needs to be also seen by a specialist as indicated by the answer to one or more of the questions below
 - *Tick boxes are provided alongside the key words to help you visually identify any problems or areas of potential concern, and provide documentation for the patient file*

We hope this leaflet will assist you in achieving successful, patient-focused interviews. The order of the questions below should be dictated by the needs of the patient.



GENERAL

The following general open-ended questions will provide an overview that may guide you immediately to specific areas of potential concern. Allow your patients to do most of the talking; demonstrate your understanding by reflecting back what they have told you, clarify when necessary. Try to avoid making the patient feel rushed, even if your time is limited.

- Have you experienced any new symptoms since we last spoke?
- How are you feeling generally?
- What do you feel has been going well?
- How active are you? Do you feel increasing fatigue when taking exercise?
- How are you sleeping?

NOTES



MEDICATIONS

The patient's white blood cell cystine level should be tested regularly (at least twice/year) and maintained at trough levels of <math><1\text{nmol hemicystine/mg protein}</math>. Trough level measurements to be taken 6 hours after 4 x times daily cystine-depleting therapy and 12.5 hours after 2 x times daily cystine-depleting therapy. If the levels are elevated check dosing (maximum dose of \geq 5\%) are vomiting, nausea, abdominal pain, breath odour, diarrhoea, skin odour, fatigue, rash and headache.

- Current dose of cystine-depleting therapy
- Adhering to cystine-depleting therapy YES NO
- Any new complaints YES NO
- Are side effects manageable? YES NO
- Missing doses?
If so, how often? YES NO
- Challenges with dosing regimen YES NO
- Reasons for missing doses; forget, or unable to for practical reasons?
- What about other medications; managing to take them? YES NO
 - Please list other medications

Find a practical solution to help your patient maintain regular dosing.



RENAL SYSTEM

SPECIFIC FOR PATIENTS WITH NATIVE KIDNEYS

- Still taking cystine-depleting therapy YES NO
- Adherence to general treatment of Fanconi YES NO

SPECIFIC FOR PATIENT ON DIALYSIS

Maintenance dialysis may be needed before kidney transplantation. Check the following with your patient while they are waiting for a kidney graft.

- Persistent polyuria YES NO
- Still taking cystine-depleting therapy YES NO
- Persistent need for electrolytes supplementation (potassium, phosphate, bicarbonates)
YES NO

SPECIFIC FOR PATIENT POST KIDNEY TRANSPLANT

- Any variation in residual urine volume YES NO
- Persistent need for electrolytes supplementation (potassium, phosphate, bicarbonates)
YES NO
- Since transplant, patient has resumed/continued to take cystine-depleting therapy
YES NO
- Adherence to immunosuppression therapies YES NO
- Other concerns? YES NO
 - *Consider referral to back to transplant team*



EXTRARENAL SYSTEM

OCULAR INVOLVEMENT

- Any reduced vision noted YES NO
- Increased sensitivity to bright sunlight (photophobia) noted YES NO
- Visual quality affected; (halos, peripheral flashing lights etc...) noted YES NO

- Compliance with eye drops regimen YES NO
 - (check technique)
- Any side effects once eye drops instilled (irritation, redness, visual blur or foreign body sensation etc.) YES NO
- Date of last review with ophthalmologist
 - Consider referral to specialist ophthalmologist

DENTAL

- Recent toothache YES NO
- Any soreness in mouth YES NO
- Bleeding gums YES NO
- Bad smell or peculiar taste in mouth YES NO
- Burning tongue or reduced taste perception YES NO
- Insufficient saliva to swallow food YES NO
 - Date last seen by a dentist
 - Consider referral to specialist dentist

GASTROINTESTINAL AND DIET

- Eating and drinking normally YES NO
- Any issues with appetite YES NO
 - Any weight loss noted YES NO
Consider referral to dietician
 - Any digestive symptoms (e.g. heartburn, reflux) YES NO
Review medications; consider referral to specialist dietician if appropriate

MUSCLE INVOLVEMENT

- Changes in muscle strength (mainly distal muscles in the 4 limbs) noted? YES NO
- Changes in muscle wasting (mainly distal muscles in the 4 limbs) noted? YES NO
- Dysphagia noted? YES NO

- Voice changes noted? YES NO
- Coughing/aspiration during feeding? YES NO
- Tires more easily during physical activity? YES NO
- Changes in handwriting (*check if they find fine motor skills more difficult*) YES NO
- Able to perform fine motor tasks [*insert a fine dexterity task appropriate for the patient, e.g. instrument playing, drawing, handicraft, etc*] YES NO



NEUROLOGICAL INVOLVEMENT

- Any headache noted? YES NO
- Any complaints about attention deficit, memory deficit, or cognitive changes YES NO
- Any gait difficulties (imbalance, stiff gait, slow gait...) noted? YES NO
- Any neurological features (motor or sensory deficit, tremor, seizures)? YES NO

RESPIRATORY

- Breathing difficulties noted when at rest and/or during exercise YES NO



ENDOCRINE INVOLVEMENT

- Monitor for diabetes mellitus (especially if patient is post-kidney transplant) YES NO
 - Test for hypothyroidism (total thyroxine and thyroid-stimulating hormone thyroid function tests) YES NO
 - Test for hypogonadism YES NO
- Ask about emergence of secondary sexual characteristics in adolescence (*if appropriate*)
- *Consider referral to endocrinologist if concerns for any of the above*

FAMILY PLANNING

- Wish to have children YES NO
- Ask if would like to receive genetic counselling YES NO
 - *Consider referral to genetic counsellor*
 - *In pregnancy, consider referral to specialist team of obstetrics and gynaecology*

- Still taking cystine-depleting therapy whilst planning pregnancy YES NO
- *[If pregnant or wishing to become pregnant]* Check understanding of when to stop and restart cystine-depleting therapy? YES NO

DERMATOLOGY

- Suggest and advise high Sun Protection Factor (>30) sunblock (*as required*)
- Any skin changes noted YES NO
 - *Note if skin more sensitive, irritable, dry etc.*

For post-transplant patients

- Any skin changes noted since transplant YES NO
 - *Consider referral back to dermatologist and communicate with transplant team*



PSYCHOLOGICAL/SOCIAL ASPECTS

- Concerns about mood (ask family/carer?) YES NO
- Financial difficulties/worries YES NO
- Concerns with school/university/college/social life YES NO
- Concerns at work YES NO
- Issues with relationship (with spouse/partner) YES NO
- Family coping? Would like additional support YES NO
- Attended patient support groups YES NO
- Would like to be put in touch with any other patients affected by cystinosis YES NO
- Name of primary point of contact within care team YES NO
- Requires additional information YES NO
 - *Ask permission to provide new information; what and how*
- Any questions/concerns not already covered YES NO
 - *Consider referral to specialist psychologist*

ADDITIONAL NOTES

Background

This checklist has been compiled by a steering committee of clinicians from across Europe with expertise in the treatment and management of patients with cystinosis throughout all stages of their life. The checklist aims to support healthcare professionals in their daily clinical practice by providing a focus to guide regular consultation with their cystinosis patients. The list of suggested key words is designed to prompt the consulting physician to ask a series of questions that will provide an overall assessment of the patients' well-being.

The key words are specifically designed to allow healthcare professionals to follow structured consultations and cover all aspects of the multi-organ effects of cystinosis, specifically with older teenagers and adult patients with cystinosis. The physician should ask open questions regarding exploratory points of interest, followed by an incentive for follow-up questions that force the patient/caregiver to think about the potential consequences and solutions to problems. When asking the patient these questions, the consulting physician should consider whether the answer provided by the patient raises a concern that merits an immediate referral to an appropriate specialist.

The expert steering committee were supported by a multidisciplinary expert faculty with expertise in the long-term support of patients with cystinosis; members of each group are given below:

Steering committee

Professor Nicholas Webb (Professor of Paediatric Nephrology, UK)
Dr Aude Servais (Nephrology and Transplant Specialist, France)
Professor Anibh Das (Professor of Paediatric Metabolic Medicine, Germany)
Dr Sally Hulton (Consultant Paediatric Nephrology, UK)
Professor Elena Levchenko (Professor of Paediatrics, Belgium)
Professor Christoph Wanner (Professor of Nephrology, Germany)

Expert faculty

Dr Gema Ariceta (Consultant Paediatric Nephrology, Spain)
Dr Hong Liang (Consultant Ophthalmology, France)
Professor Karin Lange (Professor of Medical Psychology, Germany)
Professor Francesco Emma (Professor of Paediatric Nephrology, Italy)
Professor Rezan Topaloglu (Professor of Paediatric Nephrology, Turkey)
Dr Rebecca Sberro-Soussan (Consultant Transplant Nephrology, France)
Dr David Game (Consultant Adult Nephrology, UK)
Dr Risto Lapatto (Consultant Metabolic Disease, Finland)