

**First round of consultation to incorporate patients' perspective in the development of a clinical practice guideline for inherited retinal dystrophies**

Select your diagnostic:

- ☐ Retinitis Pigmentosa
- ☐ Usher's Syndrome
- ☐ Bardet-Biedl Syndrome
- ☐ Laurence-Moon Syndrome
- ☐ Choroideremia
- ☐ Stargardt's Disease
- ☐ Atrophy Girata
- ☐ Other

Age:

Sex:

- ☐ Man
- ☐ Woman

Autonomous Community of residence:

- ☐ Andalusia
- ☐ Aragon
- ☐ Asturias
- ☐ Balearic Islands
- ☐ The Canary Islands
- ☐ Cantabria
- ☐ Castilla - La Mancha
- ☐ Castilla and Leon
- ☐ Catalonia
- ☐ Valencia
- ☐ Extremadura
- ☐ Galicia
- ☐ The Rioja
- ☐ Community of Madrid
- ☐ Navarre
- ☐ Basque Country
- ☐ Murcia Region
- ☐ Autonomous City of Ceuta
- ☐ Autonomous City of Melilla

1. What are the main disease-related health problems? Please, explain the reason for its importance.
2. What are the most common disease-related unmet needs and reasons for dissatisfaction with the Spanish NHS?

3. Do you consider appropriate that the CPG includes recommendations for additional treatments alternative to commonly used drugs for the disease? Which ones? Please, explain the reason in each case
4. How much time elapsed from first symptoms to final diagnosis?
5. How many years have passed since you were diagnosed?
6. How was the disease diagnosed?
  - ☐ Electroretinogram
  - ☐ Genetic Diagnosis
  - ☐ Other
7. Degree of disability associated with the disease (%)
8. What is your current treatment?
9. Do you currently need assistance from friends, family or carers?
10. Do you currently receive or have received visual rehabilitation?
  - ☐ Yes
  - ☐ No
11. With the result you are:
  - ☐ Strongly agree
  - ☐ Agree
  - ☐ Disagree
  - ☐ Strongly disagree
12. Do you currently receive or have received psychological support?
  - ☐ Yes
  - ☐ No
13. With the result you are:
  - ☐ Strongly agree
  - ☐ Agree
  - ☐ Disagree
  - ☐ Strongly disagree

**Second round of consultation to incorporate patients' perspective in the development of a clinical practice guideline for inherited retinal dystrophies**

Here are the 10 most important disease-related health problems identified by the 98 participants in the first round of consultation.

1. What is the importance of these problems for you? Please rank each of the following items, from 1 (not important item) to 10 (extremely important item).

	1	2	3	4	5	6	7	8	9	10
1. Lack of autonomy for daily life activities										
2. Night blindness										
3. Reduced visual field										
4. Progressive loss of visual acuity										
5. Abnormal sensitivity to light or glare										
6. Reduction of visual contrasts and colour recognition										
7. Falls										
8. Headache and / or neck contracture to compensate for visual impairment										
9. Loss of vision in relief										
10. Eyestrain										

2. Please rank the following items in order of importance for you, assigning 1 to the least important item and 10 to the most important item.

1. Lack of autonomy for daily life activities	
2. Night blindness	
3. Reduced visual field	
4. Progressive loss of visual acuity	
5. Abnormal sensitivity to light or glare	
6. Reduction of visual contrasts and colour recognition	

7. Falls

8. Headache and / or neck contracture to compensate for visual impairment

9. Loss of vision in relief

10. Eyestrain


Here are the 9 most common disease-related unmet needs and reasons for dissatisfaction with the Spanish NHS identified by the 98 participants in the first round of consultation.

3. What is the importance of these reasons for dissatisfaction for you? Please rank each of the following items, from 1 (not important item) to 10 (extremely important item).

1 2 3 4 5 6 7 8 9 10

1. Lack of valid and homogeneous protocols for diagnosis and clinical management

2. Waiting lists for diagnostic tests and monitoring

3. Lack of specialized care units

4. Scarcity of research on therapeutic options

5. Lack of professionals empathy

6. Lack of support in the adaptation process (professional, technical and economic)

7. Access difficulties to genetic studies

8. Access difficulties to treatment or counseling

9. Lack of information about the disease


4. Please rank the following items in order of importance for you, assigning 1 to the least important item and 9 to the most important item.

1. Lack of valid and homogeneous protocols for diagnosis and clinical management

2. Waiting lists for diagnostic tests and monitoring

3. Lack of specialized care units


4. Scarcity of research on therapeutic options

5. Lack of professionals empathy

6. Lack of support in the adaptation process (professional, technical and economic)

7. Access difficulties to genetic studies

8. Access difficulties to treatment or counseling

9. Lack of information about the disease


Here are the 6 most relevant disease-related treatment aspects identified by the 98 participants in the first round of consultation.

5. What is the importance of these treatment aspects for you? Please rank each of the following items, from 1 (not important item) to 10 (extremely important item).

1 2 3 4 5 6 7 8 9 10

1. Adequate protective and aids devices

2. Comprehensive rehabilitation (visual and / or auditory)

3. Recommendations to prevent worsening

4. Healthy behaviors: diet, physical activity, adequate rest and activities to avoid

5. Psychological support

6. Vitamins and other supplements


6. Please rank each of the following items in order of importance for you, assigning 1 to the least important item and 6 to the most important item.

1. Adequate protective and aids devices

2. Comprehensive rehabilitation (visual and / or auditory)

3. Recommendations to prevent worsening

4. Healthy behaviors: diet, physical activity, adequate rest and activities to avoid

5. Psychological support


6. Vitamins and other supplements



### **Third round of consultation to incorporate patients' perspective in the development of a clinical practice guideline for inherited retinal dystrophies**

Here are the 10 most important disease-related health problems in order of importance after two rounds of consultation.

1. What is the importance of these problems for you? Please rank the following items in order of importance, assigning 1 to the least important item and 10 to the most important item. Our aim is generate as much agreement as possible among participants.

1. Lack of autonomy for daily life activities
2. Night blindness
3. Reduced visual field
4. Progressive loss of visual acuity
5. Abnormal sensitivity to light or glare
6. Reduction of visual contrasts and colour recognition
7. Falls
8. Headache and / or neck contracture to compensate for visual impairment
9. Loss of vision in relief
10. Eyestrain


Here are the 9 most common disease-related unmet needs and reasons for dissatisfaction with the Spanish NHS in order of importance after two rounds of consultation.

2. What is the importance of these reasons for dissatisfaction for you? Please rank the following items in order of importance, assigning 1 to the least important item and 9 to the most important item. Our aim is generate as much agreement as possible among participants.

1. Lack of valid and homogeneous protocols for diagnosis and clinical management
2. Waiting lists for diagnostic tests and monitoring
3. Lack of specialized care units


4. Scarcity of research on therapeutic options
5. Lack of professionals empathy
6. Lack of support in the adaptation process (professional, technical and economic)
7. Access difficulties to genetic studies
8. Access difficulties to treatment or counseling
9. Lack of information about the disease


Here are the 6 most relevant disease-related treatment aspects identified in order of importance after two rounds of consultation.

3. What is the importance of these treatment aspects for you? Please rank the following items in order of importance, assigning 1 to the least important item and 6 to the most important item. Our aim is generate as much agreement as possible among participants.

1. Adequate protective and aids devices
2. Comprehensive rehabilitation (visual and / or auditory)
3. Recommendations to prevent worsening
4. Healthy behaviors: diet, physical activity, adequate rest and activities to avoid
5. Psychological support
6. Vitamins and other supplements


Finally,

Do you have any suggestions to future research on inherited retinal dystrophies?