Co-funded by
the European Union

seeing the invisible

DETECTION OF ANIRIDIA

OERs ADAPTED TO STUDENTS WITH LOW VISION.

redREE making projects

ALBA Asociación de ayuda a personas con albinismo

Aniridia Italiana

ANIRIDI NORGE

ANIRIDIA EUROPE

smallcodes

académie Clermont-Ferrand greta du VELAY

Coordinator entity: RedTree Making Projects Coop.V.

Address: Jesús y María 26 - ground floor.
46008 - Valencia, Spain.

e-mail: info@redtree.es

Phone: 96 015 06 04

This result has been developed by Redtree Making Projects Coop. V. in collaboration with GRETA du Velay, Smallcodes, Aniridia Europe, Alba Asociación, Aniridia Norway and Aniridia Italiana within the project "SEEING THE INVISIBLE: Inclusive digitalization of low vision students in school education", cofinanced by the ERASMUS+ PROGRAMME of the EUROPEAN UNION.

This project has been funded with the support of the European Commission.
The author is exclusively responsible for this publication.
The Commission cannot held liable for any use of the information herein.

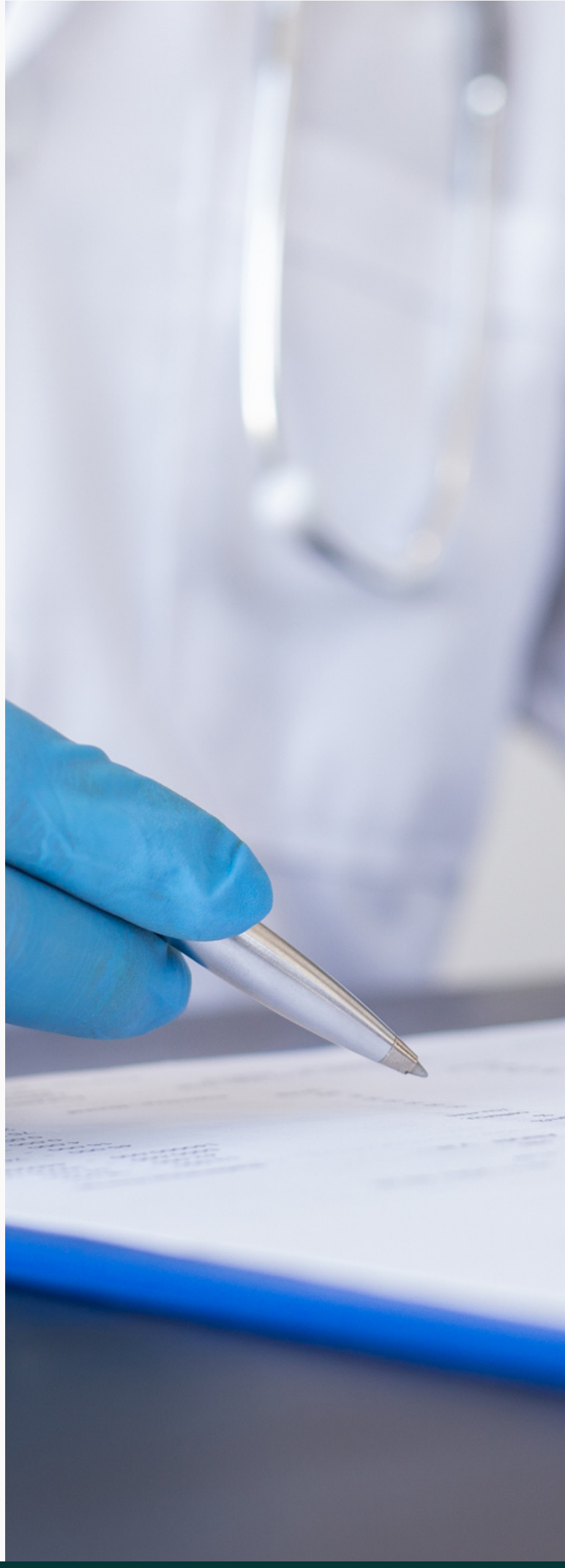
Co-funded by the European Union

seeing the invisible

CONTENTS

I. INTRODUCTION

Aniridia is a rare genetic eye disorder. Its prevalence is estimated at one in every 40.000 to 100.000 births[1]. Its derived problems usually occur from birth, although others may appear later both in childhood, adolescence, and later.



II. SYMPTOMS OF ANIRIDIA

- Iris hypoplasia: very large pupils → photophobia.
- Macular, foveal, and optic nerve hypoplasia. Lens subluxation → decreased visual acuity, nystagmus, strabismus, amblyopia.
- Problems in drainage of the eye → dry eye, glaucoma, eye pain, corneal degeneration.
- Refraction problems → myopia, hyperopia, astigmatism.

Possible linked complications:

- Glaucoma, cataracts, keratopathy, retinal detachment... can be developed later.
- WAGR syndrome: acronym for Wilms tumor (risk of developing a kidney tumor up to 10 years old –in 40 to 70% of cases–), Aniridia, Genitourinary anomalies, and Retardation in intellectual development.
- t can also be coupled with other conditions like olfactory reduction, hearing loss, sleep pattern alterations, or endocrine alterations.
- Cerebellar ataxia or Gillespie syndrome is related to Aniridia, although it is extremely rare. It is characterized by a psychomotor and intellectual maturity delay, and lack of coordination of muscle movements due to developmental problems of the cerebellum.

III. DETECTION OF ANIRIDIA

The diagnosis is relatively simple, since the partial or total lack of the iris in the eye can be seen at first glance. However, it could be confused with an iris coloboma, which is a different disease caused by a congenital problem that creates a fissure in the iris or a lack of part of it, without necessarily involving a serious impairment of visual acuity or derived problems like glaucoma or cataract.

Aniridia is usually diagnosed at birth or in the first years of life, with both ophthalmological tests and examinations and genetic diagnoses to confirm the type of the disease, facilitating the detection of possible future complications.

A child with Aniridia will show an obvious photophobia, closing his eyes to light sources, preferring to be in the shadows.

Other ocular and visual conditions, and others related to the WAGR syndrome can also be added to the lack or condition of the iris. All these related genetic conditions have Aniridia in common.

Sometimes the condition is detected even before birth, thanks to ultrasounds with which certain alterations to the genitourinary system can be verified, although it is usually detected in neonatal examination due to ocular and/or these genitourinary alterations.

IV. TREATMENT

The treatments applied in cases of Aniridia are aimed at alleviating or correcting the alterations and effects it produces.

- Regular eye tests to monitor vision.
- Early visual stimulation to develop the remaining sight.
- Glasses of different types (sunglasses, graduated glasses, “pinholes”...) to protect the eye from light and/or correct both visual acuity and refractive problems.
- Eye patches for amblyopia.
- Drugs for various symptoms, like drops to treat dry eye.
- Visual aids (magnifying glasses, mini-telescope, tele-magnifying glasses, etc.).
- Low vision contact lenses can be used to make up for the function of the iris, regulating the input of light, thus improving sight and the external appearance of the eye.
- Microsurgery is a possible option for iris reconstruction or to replace it with an artificial one. Surgery is also used to correct other problems, like cataracts, nystagmus, or trabeculectomy to treat glaucoma. Transplants may also be used for some treatments, like limbus to treat corneal degeneration.
- Flexible and personalized artificial irises are currently being developed, which can be implanted after cataract surgery.
- Ultrasounds, scans, or magnetic resonance imaging to detect the appearance of Wilms tumor early, and to treat it with chemotherapy and/or surgery if it appears.

V. RECOMMENDATIONS

1. Explain to the child that they have a very large pupil, which is why light hurts them, and that they should use protective (sunglasses) and corrective measures where appropriate.

2. Explain things to them naturally, without dramatizing and telling the truth.

3. Encourage them to make a life as similar as possible to that of other children their age.

4. Provide them with the necessary support to avoid the appearance of complexes and disabling fears.

5. Make them feel fully integrated while strengthening their self-awareness in the normal use of prescribed and necessary corrective and supportive measures.

Co-funded by
the European Union

seeing the
invisible

redREE
making projects

ALBA
Asociación
de ayuda a
personas con
albinismo

Aniridia Italiana

ANIRIDI
NORGE

ANIRIDIA EUROPE

smallcodes

académie
Clermont-Ferrand
greta
du VELAY