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New rare disease European Network led by Sweden

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Aniridia is a rare congenital eye disease, affecting about one in 80,000 people, and requiring intensive eye care, social and community support from birth and throughout an individual's lifetime. Most ophthalmologists will have come across one or more cases of aniridia during their careers. Although classified as a rare disease, congenital aniridia is a pan-ocular disease characterized by an underdevelopment or improper development of eye structures, often leading to iris and foveal hypoplasia, nystagmus, cataract, glaucoma, limbal stem cell insufficiency, keratopathy and dry eye. For this reason, despite its rarity, aniridia requires significant eye care throughout life, and many subspecialties will have contact with persons with aniridia. Congenital aniridia is caused primarily by a mutation in the PAX6 gene responsible

for ocular development *in utero*, however, over 500 unique mutations in this gene have been reported, resulting in a range of ocular phenotypes and prognoses. This complexity, combined with the rarity of the disease, means that very few effective treatments are available. What is needed is adequate-sized patient populations to conduct coordinated clinical and research activities, and improved information exchange in assessing and treating aniridia, with expertise being shared across geographically dispersed centers.

ANIRIDIA-NET (www.aniridia-net.eu) is a pan-European bottom-up network of researchers, ophthalmologists, trainees, aniridia patient organizations, industry and special interest groups funded by the European Union's Cooperation in Science and Technology (COST) program

(www.cost.eu) for the period May 2019 – April 2023. The network is chaired by Prof. Neil Lagali in Linköping, Sweden with Vice-Chair Prof. Claus Cursiefen from Cologne, Germany. ANIRIDIA-NET currently consists of 28 European member countries and 2 near neighbor countries (Figure 1), with all the Nordic Countries represented. The overall goal of ANIRIDIA-NET is to improve clinical management of aniridia and promote innovative research and development of new alternatives for its diagnosis and treatment. This network encourages communication among healthcare professionals, researchers, and patient representatives to share their ideas and foster collaboration to improve aniridia management through evidence-based research, harmonized clinical protocols, pooling/sharing of samples and models and consensus activities.

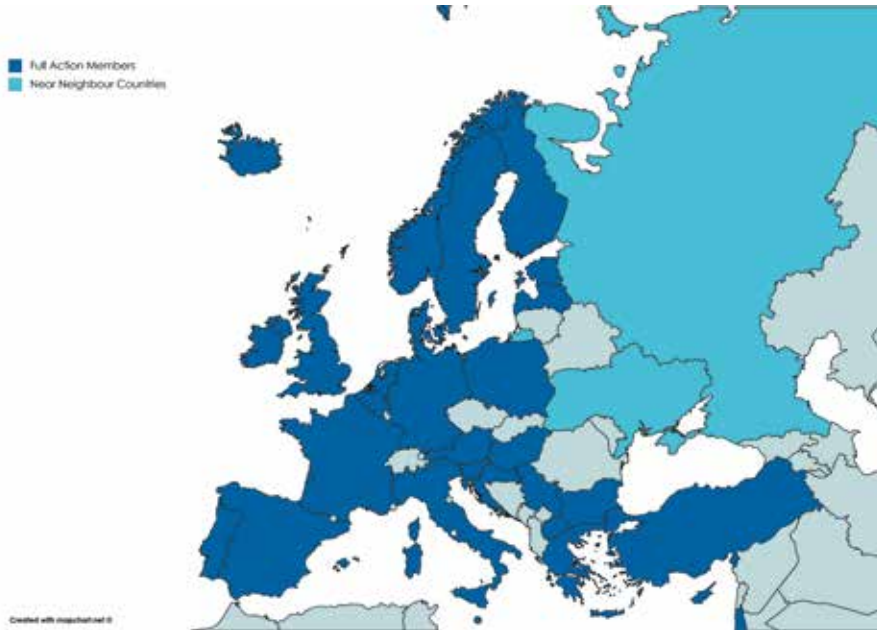


Figure 1. Geographical distribution of ANIRIDIA-NET members, including 30 participating countries, 28 COST full or cooperating members (darker blue) and 2 near neighbour countries (lighter blue). Date updated: 14/04/2020. Created with mapchart.net.

ANIRIDIA-NET will achieve its goals through directed tasks and activities aligned along six different themes, each being the responsibility of a specific working group:

- (1) **Clinical guidelines.** Harmonization/consensus on clinical examinations, treatment guidelines, patient information and clinical endpoints.
- (2) **Clinical and cohort studies.** Development of clinical and cohort multicenter studies with patient data, and/or biological samples for biomarker, genetic and high-throughput molecular analysis.
- (3) **Stem cells and regenerative medicine.** Approaches of stem cell research, tissue engineering, genetic techniques for developing translational regenerative therapeutic strategies.
- (4) **Transplantation, inflammation and immunity.** Evaluation of emerging surgical techniques for aniridia associated pathologies.
- (5) **Aniridia models for collaborative research.** Development of aniridia animal models and novel research techniques for the identification of new therapeutic strategies.
- (6) **Patient-driven research.** Support of patient participation in aniridia-focused research to raise awareness toward unmet patient needs and challenges.

The ANIRIDIA-NET consortium had a first kickoff meeting in Brussels in April 2019 (Figure 2), followed by a further meeting in Paris in September 2019, and a third meeting in Lisbon in late February 2020 (Figure 3). In June 2019, ANIRIDIA-NET co-organized a Nordic Countries Aniridia Conference held at the Hurdal vision rehabilitation center on a picturesque lake north of Oslo. A panel of expert researchers, clinicians and a geneticist presented information to patients and answered their questions in lively discussions. Additionally, ANIRIDIA-NET supports conference symposia such as a Special Interest Session on aniridia held at the European Vision and Eye Research annual congress in Nice in October 2019, where investigators across Europe presented their latest findings in aniridia-related research.



Figure 2. ANIRIDIA-NET members at the Action's kick-off meeting held in Brussels on April 2019. Networking meetings serve as essential support for collaboration, knowledge sharing and research promotion.



Figure 3. ANIRIDIA-NET members at the 3rd scientific meeting held in Lisbon in February 2019, coinciding with Rare Disease Day. Culture and assessment of limbal stem cells, clinical multicentre studies, harmonized clinical protocols, aniridia pathophysiology, post-transplant immunosuppression and MSCs for eye diseases were some of the topics covered during this networking event.

Training is an important component of ANIRIDIA-NET, and in 2020-2021, two Training Schools will be organized, and these are open to doctoral, postdoctoral and resident trainees across Europe. Training schools combine lectures given by experts with hands-on practical wetlabs, workshops and social activities to foster networking among the trainees. Another important feature of this COST Action is the possibility for researchers or clinicians to obtain funding for cross-border research within the Short-Term Scientific Mission (STSM) scheme. Collaborative projects of scientific or clinical relevance, along with training and capacity-building, can be established through the exchanges of trainees or investigators in STSMs (Figure 4).

Any individual interested in aniridia research in the Nordic Countries is welcome to join and participate in ANIRIDIA-NET activities. This network is an open and inclusive group following EU-mandated principles. For more information and contact points for each working group, please visit the website www.aniridia-net.eu.



Figure 4. Elena Danielle, a MSC postgraduate at the Veneto Eye Bank Foundation (Italy), during a Short Term Scientific Mission (STSM) at the research group led by Heli Skottman at Tampere University (Finland). The goal of this STSM was to share knowledge on primary limbal stem cell culture and characterization techniques. STSMs give researchers the opportunity to foster linkages and build capacity in research.



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