



# GENETICALLY DEFINED EDS:

Strategies & Solutions for Unmet Needs

AUGUST 30-31, 2023

**BELGIUM**



**EDS ECHO SUMMIT SERIES**

## **GENETICALLY DEFINED EDS:** Strategies & Solutions for Unmet Needs

**PRESENTATION**

Organisation of Care for Ehlers-Danlos  
syndromes in the UK

**SPEAKER**

**Fleur S. van Dijk & Glenda Sobey**  
Lead National EDS service London & Sheffield

## **GENETICALLY DEFINED EDS:**

Strategies & Solutions for Unmet Needs

AUGUST 30-31, 2023

**BELGIUM**



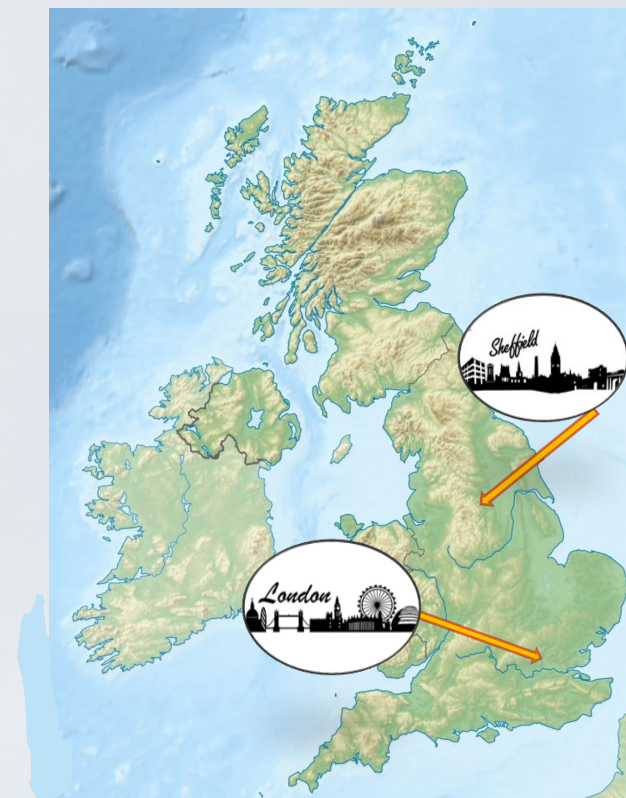
### **National EDS service London and Sheffield**

**Highly specialized service within National Health Service (NHS) since 2009**

**Clinical Team of EDS consultants and counsellors**

**Diagnostic service for rare, monogenic EDS types**

**Follow-up of rare, monogenic EDS types**



### **Referrals:**

- Suspicion on a rare, monogenic type of EDS or confirmed diagnosis of a rare, monogenic type of EDS
- From NHS specialists in secondary and tertiary care for children and adults

### **Pre-clinic**

- Before their clinical appointment with the consultant, patients will be contacted by an EDS counsellor for clinical information and family history

### **Clinic**

- Patients are seen by an EDS consultant for clinical assessment and if indicated, additional investigations including genetic testing

### **Post-clinic**

- Patients are informed about results of additional investigations.



# **GENETICALLY DEFINED EDS:**

Strategies & Solutions for Unmet Needs

AUGUST 30-31, 2023

**BELGIUM**





# GENETICALLY DEFINED EDS:

Strategies & Solutions for Unmet Needs

AUGUST 30-31, 2023

BELGIUM



**What happens after a diagnosis of a rare type of EDS?**







Provide information for clinicians and address questions

- Information for patients & families (leaflets, emergency and care cards)
- Available for further questions & advice
- Genetic counselling support



Cardiovascular surveillance  
(joint clinics with cardiologist and vascular surgeon)



Clinical research  
(neEDS study: natural history exploration of rare **EDS**)

Patient charities



Referrals to other specialties  
e.g. rheumatology, gastroenterology, hematology...

# Strength in Numbers



Contribute to knowledge internationally to improve care for people with genetically defined EDS types



# GENETICALLY DEFINED EDS:

Strategies & Solutions for Unmet Needs

AUGUST 30-31, 2023

**BELGIUM**



Thank you  
for listening

