



International Summer School on Rare Disease Registries and FAIRification of Data

September 27 – October 1, 2021 Istituto Superiore di Sanità, Rome, Italy

Endorsed by





GENERAL INFORMATION

Due to the force majeure situation and in order to allow the correct progress of the planned tasks of EJP RD, ISS ensures that the international course "International Summer School on Rare Disease Registries and FAIRification of Data", September 27 - October 1, 2021, ISS, Rome will be held ONLINE.

RELEVANCE, INTRODUCTION AND OBJECTIVES

The International Summer School on Rare Disease Registries and FAIRification of Data is a part of a series of training activities proposed by the European Joint Programme on Rare Diseases (EJP RD). EJP RD is a European Commission funded project (grant agreement No 825575, 2019 – 2023) with the goal "to create a comprehensive, sustainable ecosystem allowing a virtuous circle between research, care and medical innovation". For more information about the EJP RD, see https://www.ejprarediseases.org/

This Course is a part of the WP14 on "Data Management & Quality Training", which aims to organize residential training courses in different Countries. WP and Task Leader: Dr. Claudio Carta, ISS.

Course Director: Dr. Domenica Taruscio, ISS.

The Course is made up of 5 days of training organised by Istituto Superiore di Sanità (ISS) in close collaboration with, mainly, EJP RD task partners [LUMC & UoG (Endo-ERN), IOR (Bond-ERN), UUH (Metab-ERN), EURORDIS, ISCIII, LUMC, INSERM (RaDiCo), UMCG, DTL-Projects (ELIXIR-NL), CNR (ELIXIR-IT), AMC]

The Course is endorsed by the International Conference On Rare Diseases and Orphan Drugs (ICORD).

ISS, has gained vast experience by organising numerous courses focused on rare disease registries with the support of key partners. Since 2013 ISS has organised and hosted the "International Summer School on Rare Disease and Orphan Drug Registries" and since 2014 the "Bring Your Own Data To Link Rare Disease Registries".

SPECIFIC LEARNING OBJECTIVES

Registries are key resources to increase timely and accurate diagnosis, improve patient's management, tailor treatments, facilitate clinical trials, support healthcare planning and speed up research.

This course is composed of two training modules:

- During the first three days module (27-29 September 2021), participants will learn (a) what resources are needed for the establishment/maintenance of a high-quality registry (b) the features of successful strategies to ensure (i) long-time sustainability of the registry, (ii) quality, (iii) legal and ethical issues in compliance with the EU (European Union) General Data Protection Regulation.
- During the second two days module "FAIRification of data", (September 30 October 1, 2021) participants will deepen their knowledge on the single steps of the FAIRification of data and will discover the potential of FAIR registries. In this part a time slot will be allocated to discuss FAIR data management and FAIR project planning.



At the end of the training course participants will be able to:

- Identify the methodologies and appropriate characteristics to realize and run a rare disease registry
- Describe how to collect quality data
- List the main steps of the FAIRification process
- Describe the main features of a FAIR registry

LEARNING METHOD

In the first and second module there will be interactive plenary presentations and question & answer sessions between speakers and participants. Moreover in the second module there will be hands-on exercises and a wrap up session.

PARTICIPANTS AND REGISTRATION

The training course is open to the international research community, clinicians, medical specialists, registry curators, database managers, healthcare professionals and rare disease patients' representatives.

To ensure active participation and exchange with teaching staff and participants, a maximum of 30 attendees will be admitted to each training module. A selection process will be applied based on the participants' background and role with reference to registry activities, and on the involvement in ERNs.

REGISTRATION

Registration is possible for:

- > the first training module: "Rare Disease Registries", September 27-29, 2021
- > the second training module: "FAIRification of Data", September 30-October 1, 2021
- > the entire course: "Rare Disease Registries" and "FAIRification of Data", September 27 October 1, 2021.

REGISTRATION FORM For important updates, deadlines and for the online registration please visit the website at the following LINK

FEES AND COSTS

The course and registration are free of charge.

The course organisers will not cover expenses incurred by the participants in any case.

LEARNING ASSESSMENT

At the end of each training module participants will be asked to submit a learning assessment, based on an online multiple-choice questionnaire and a satisfaction survey.

ATTENDANCE CERTIFICATES

At the end of the course a certificate of attendance will be forwarded to the participants who attended 100% of the single training modules or the entire course program. No credits of Continuing Education in Medicine will be issued.



OFFICIAL LANGUAGE

English

VENUE

ONLINE

Important dates, deadlines, registration form, and further information, please visit the website at the following LINK

CONTACT

If you have questions, please write to the cou<mark>rse Coordinator Dr Claudio Carta: claudio.carta@iss.it (in Cc laura.cellai@iss.it)</mark>



Programme of the Course

DAY 1 (CEST Time) (LINK)

1st Training Module, September 27, 2021

https://teams.microsoft.com/l/meetup-

join/19%3ameeting MWY4MDYxNzYtZjBkYS00MWQ4LWI1YjYtYjEyYWlwYzJjMjlj%40thread.v2/0?context=%7b%22Tid%22%3a%22383ac401-a21d-426b-8072-

5f74c36b7093%22%2c%22Oid%22%3a%226293aec5-7106-4e7a-87e1-6bed39dd917c%22%7d

14:00 Welcome address & Faculty & Presentation of the course

Domenica Taruscio

14:15 Presentation of the European Joint Programme on Rare Diseases

Claudio Carta, Domenica Taruscio

14:30 The European Platform on Rare Disease Registration (EU RD Platform) and JRC Activities

Andri Papadopoulou

15:00 Questions & Answers

15:15 Break

15:45 Aims, Governance & Sustainability

Joseph Giuliano

16:45 Questions & Answers

17:30 End of Day 1

DAY 2 (CEST Time) (LINK)

1st Training Module, September 28, 2021

https://teams.microsoft.com/l/meetup-

join/19%3ameeting ZGEyOWNkYjYtMDgxYS00OWU3LTg2Y2EtZTE0NzM5NTk4Y2Y1%40thread.v2/0?context=%7b%22Tid%22%3a%22383ac401-a21d-426b-8072-

5f74c36b7093%22%2c%22Oid%22%3a%226293aec5-7106-4e7a-87e1-6bed39dd917c%22%7d

14:00 Quality of RD Registries

Yllka Kodra

14:45 Questions & Answers

15:00 Break



15:15 Ethics, GDPR and Informed Consent

Marta Tomasi

16:15 Questions & Answers

17:00 End of Day 2

DAY 3 (CEST Time) (LINK)

1st Training Module, September 29, 2021

https://teams.microsoft.com/l/meetup-

join/19%3ameeting MmQ5NmFIZGUtYjdiNC00NTI5LWI0YjltZTdlMmY2ZDNmYjBj%40thread.v2/0?context=%7b%22Tid%22%3a%22383ac401-a21d-426b-8072-

5f74c36b7093%22%2c%22Oid%22%3a%226293aec5-7106-4e7a-87e1-6bed39dd917c%22%7d

14:00 Roles of RD patients in registries & research - ePAGs in ERNs

Gulcin Gumus

- 14:30 Questions & Answers
- 14:45 Implementing the active partnership with patients' representatives in a specific ERN Registry: the management of TogethERN ReCONNET

Diana Marinello

15:00 The perspective of patient representatives in an ERN Registry

Johan de Graaf

- 15:15 Questions & Answers
- 15:30 Break
- 15:45 RegistRare: a platform for patient registries

Tiziana Grassi, Paola Torreri

- 16:15 Questions & Answers
- 16:30 Rare disease registries and how they work in real life the EuRRECa experience

Syed Faisal Ahmed

- 17:15 Questions & Answers
- 17:45 Evaluation of the 1st Training Module and satisfaction survey
- 18:00 End of Day 3



19:00 Social Event (LINK)

https://teams.microsoft.com/l/meetup-

join/19%3ameeting_M2I5ZTU1N2UtOTAwMS00MjE0LTIjMjktYzYyYWUyOWZjMGVh%40thread.v2/0?context=%7b%22Tid%22%3a%22383ac401-a21d-426b-8072-5f74c36b7093%22%2c%22Oid%22%3a%226293aec5-7106-4e7a-87e1-6bed39dd917c%22%7d

DAY 4 (CEST Time)

2nd Training Module, September 30, 2021

MORNING SESSION (LINK)

https://teams.microsoft.com/l/meetup-

join/19%3ameeting_ZjcxNTFjNDUtMTE5Mi00M2Q3LWEwMzUtMDI1NGRiMTE2YTFl%40thread.v2/0 ?context=%7b%22Tid%22%3a%22383ac401-a21d-426b-8072-

5f74c36b7093%22%2c%22Oid%22%3a%226293aec5-7106-4e7a-87e1-6bed39dd917c%22%7d

09:50 Welcome address & Presentation of the course

Domenica Taruscio

10:00 BYOD introduction

Claudio Carta, Marco Roos

10:10 FAIR Game

Bruna Dos Santos Vieira, Marco Roos, Rajaram Kaliyaperumal, Martijn Kersloot, Alberto Cámara, César Bernabé, Clemence le Cornec, Joeri van der Velde, Shuxin Zhang

11:00 Break

11:15 Lesson Learned and the Benefits of making data FAIR

Marco Roos, Rajaram Kaliyaperumal, Bruna Dos Santos Vieira, Martijn Kersloot

12:00 The main steps of FAIRification

Marco Roos, Claudio Carta

12:10 Introduction to making data linkable and machine-readable

Mark Wilkinson

12:40 Break



AFTERNOON SESSION (LINK)

https://teams.microsoft.com/l/meetup-

join/19%3ameeting OGQwZDVmNzgtMmUxNS00YzNiLWFjZGUtOWQ1Y2UyYmZkM2Nk%40threa d.v2/0?context=%7b%22Tid%22%3a%22383ac401-a21d-426b-8072-

5f74c36b7093%22%2c%22Oid%22%3a%226293aec5-7106-4e7a-87e1-6bed39dd917c%22%7d

13:30 Describing your registry at source for machines:

- Describing your registry with 'ORDO', 'DCAT' & a 'FAIR Data Point'

Marc Hanauer, Rajaram Kaliyaperumal

- Describing your registry access protocols

Esther van Enckevort

- 14:10 Hands on (Create a FAIR Data Point)
- 14:30 Break
- 14:45 Introduction to querying a FAIR Data Point.

Mark Wilkinson

- 15:15 Hands-on Introduction to querying a FAIR Data Point
- 15:30 Using your FAIR metadata: exploring the FAIR Data Point via the Web

Rajaram Kaliyaperumal

- 15:45 Hands-on exploration of registry descriptions in a FAIR Data Point
- 16:00 Wrap-up; Questions & Answers

16:30 End of Day 4

DAY 5 (CEST Time)

2nd Training Module, October 1, 2021

MORNING SESSION (LINK)

https://teams.microsoft.com/l/meetup-

join/19%3ameeting_MTU0ZjRiNDUtY2YyZC00ZmJlLTk5OTUtMWU2NzdlMzl4OGE3%40thre

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<u>5f74c36b7093%22%2c%22Oid%22%3a%226293aec5-7106-4e7a-87e1-</u>

6bed39dd917c%22%7d

10:00 Making Data Machine-readable & Hosting FAIR data: an example of a registry becoming FAIR

Martijn Kersloot



10:20 Hands on

10:40 Wrap-up

11:00 Using ontologies to describe data unambiguously for machines (and humans)

Ronald Cornet

11:30 Break

11:45 Introducing the EJP RD ontological model for 'Common Data Elements'

Mark Wilkinson

12:00 How to apply the CDE model to your data

Automatic conversion from tabular data to machine readable data with YARRRML and Matey

Mark Wilkinson

12:30 Exploring linkable, machine readable CDEs with SPARQL: Hands-on demonstration

Mark Wilkinson

12:45 Break

AFTERNOON SESSION (LINK)

https://teams.microsoft.com/l/meetup-

join/19%3ameeting ZmRhYThmZTEtMDNmZS00Y2M1LTkyYzYtYWRmMDMyYjViMTJh%40thread.v 2/0?context=%7b%22Tid%22%3a%22383ac401-a21d-426b-8072-

5f74c36b7093<mark>%22%2c%22Oid%22%3a%2262</mark>93aec5-7106-4e7a-87e1-6bed39dd917c%22%7d

14:30 Solving the game with FAIR Data & Metadata

Bruna Dos Santos Vieira, Marco Roos, Alberto Cámara, César Bernabé, Clemence le Cornec, Joeri van der Velde, Shuxin Zhang

15:00 FAIRification recap, implications for "registry managers" and project planning

Esther van Enckevort, Marco Roos, Claudio Carta, Bruna Dos Santos Vieira

15:30 Hands on

15:50 Wrap-up

16:00 How global open FAIR data are changing the world in practice

Erik Schultes, Barend Mons

16:20 Parking lot & Q&A next steps for your own FAIR registry

16:50 Evaluation of the 2nd Training Module and satisfaction survey



17:00 Concluding remarks

Domenica Taruscio, Marco Roos, Claudio Carta

17:10 End of the Course

SPEAKERS

Syed Faisal Ahmed, University of Glasgow, (EndoERN), UK

Claudio Carta, National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy

Ronald Cornet, Academic Medical Center, Universiteit van Amsterdam, The Netherlands

Johan de Graaf, ePAG Endo ERN; president of the Dutch Pituitary Foundation, The Netherlands

Joseph Giuliano, Global Medical Operations & Patient Registries Amicus Therapeutics, USA

Tiziana Grassi, National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy

Gulcin Gumus, EURORDIS, France

Marc Hanauer, Directeur technique Orphanet, INSERM, France

Rajaram Kaliyaperumal, Leiden University Medical Centre, The Netherlands

Martijn Kersloot, Academic Medical Center, Universiteit van Amsterdam, The Netherlands

Yllka Kodra, Ministry of Health Directorate-General for Health Planning Government Organisation, Italy

Diana Marinello, Azienda Ospedaliero Universitaria Pisana, Italy

Barend Mons, Leiden University Medical Centre, GO FAIR, The Netherlands

Andri Papadopoulou, European Commission's Joint Research Centre, Italy

Marco Roos, Leiden University Medical Centre, The Netherlands

Erik Schultes, GO FAIR, The Netherlands

Domenica Taruscio, National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy

Marta Tomasi, University of Bolzano, Italy

Paola Torreri National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy

Esther van Enckevort, University Medical Centre Groeningen, The Netherlands

Mark Wilkinson, Centro de Biotecnología y Genómica de Plantas UPM-INIA (CBGP), Spain



FAIRification Stewards

César Bernabé, Leiden University Medical Centre, The Netherlands

Alberto Cámara, Centro de Biotecnología y Genómica de Plantas UPM-INIA (CBGP), Spain

Bruna dos Santos Vieira, Center for Molecular and Biomolecular Informatics, Radboud University Medical Center, The Netherlands

Clemence le Cornec, Heidelberg University Hospital, Germany

Joeri van der Velde, Groeningen University Medical Center, The Netherlands

Shuxin Zhang, Academic Medical Center, Universiteit van Amsterdam, The Netherlands

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Domenica Taruscio, National Centre for Rare Diseases, ISS, Rome, Italy

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ORGANIZING SECRETARIAT

Linda Agresta, Daniela Bernardo, Laura Lee Cellai, Patrizia Crialese, Stefano Diemoz, National Centre for Rare Diseases, ISS, Rome, Italy email: laura.cellai@iss.it



Connection Instructions & User-Guide

The International Summer School on Rare Disease Registries and FAIRification of Data is held Online using Microsoft Teams (MS Teams).

You have the choice to either:

- use your **web browser**: Google Chrome (preferably) or Microsoft Edge or Firefox (but neither Safari nor internet explorer)
- Or the MS Teams Application (recommended) if you are already using it.
 - If not, and you wish to use this application, download it at: https://www.microsoft.com/en/microsoft-365/microsoft-teams/download-app

You will need to sign in (it is free)

 Or the smartphone Teams app (available on the AppStore [iPhone] or Play Store [Android]). You will need to sign in (it is free) and you will need to open the links on your smartphone

Please Connect to the Webinar and Live Sessions at least 10 minutes before they start

Sessions

The Event will be organized as a Live Event in MS Teams. This means that you will be able to watch the speaker's video, the presentation slides and ask questions through a « Question and Answer » space (next to the video being broadcasted, see below).

The procedure to connect and interact is the following:

1/In the Agenda Click on the "LINK" of the respective session, the link will open on your "default" internet browser (or on your MS Teams desktop application if you are already using it), example below:

DAY 4 (CEST Time)

2nd Training Module, September 30, 2021

MORNING SESSION (LINK)

https://teams.microsoft.com/l/meetup-

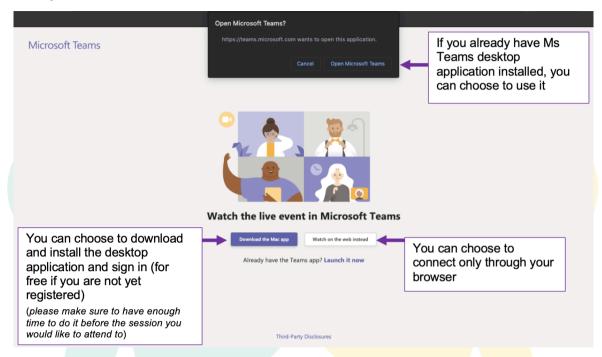
join/19%3ameeting_ZjcxNTFjNDUtMTE5Mi00M2Q3LWEwMzUtMDI1NGRiMTE2YTFl%40thread.v2/0 ?context=%7b%22Tid%22%3a%22383ac401-a21d-426b-8072-

5f74c36b7093%22%2c%22Oid%22%3a%226293aec5-7106-4e7a-87e1-6bed39dd917c%22%7d

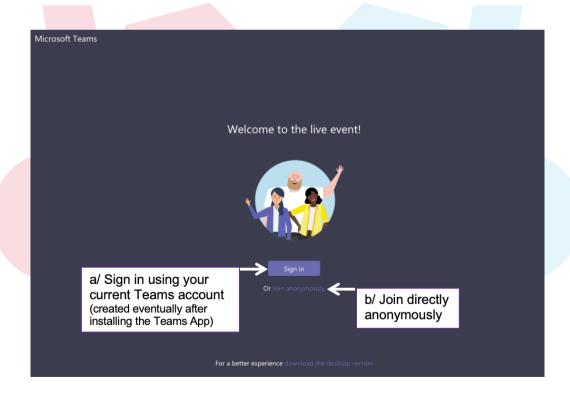
Or right click on the "LINK"; copy the link and paste it in the internet browser of your choice (Chrome, Microsoft Edge or Firefox)



2/ On your browser:

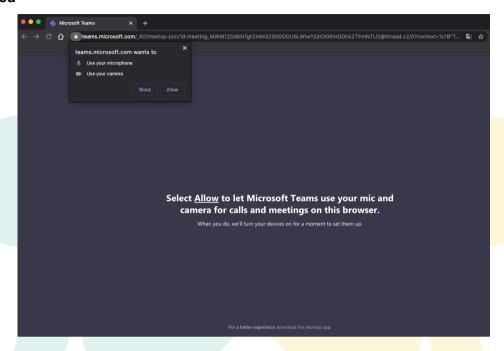


3/ Whether you choose to "Watch on the Web" or using MS Teams desktop application, if you are not signed in MS Teams you will have the choice to:



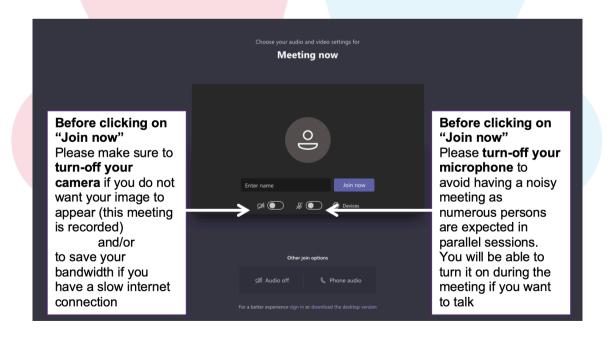


4/ If you decide to "Join on the web" you need to allow the use of "your microphone" to be able to talk and the use of "your camera" to at least be able to share your screen if needed



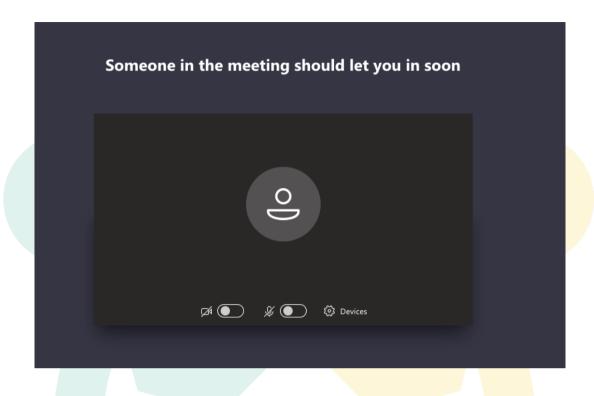
5/ Please Enter the "First name" and "Last name" you provided when you registered to be admitted to the different sessions

[If you are already using MS Teams desktop application, or you have already downloaded this application and signed in, your initials and your email will be automatically displayed. Please do not click on "Add a room"]

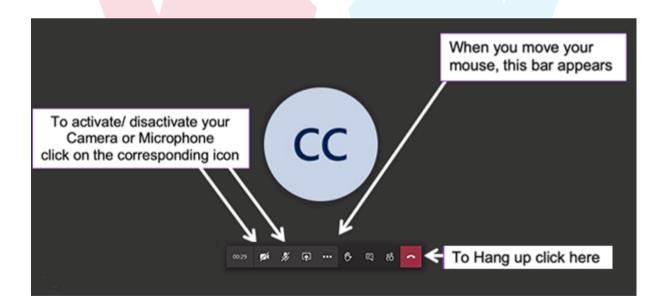




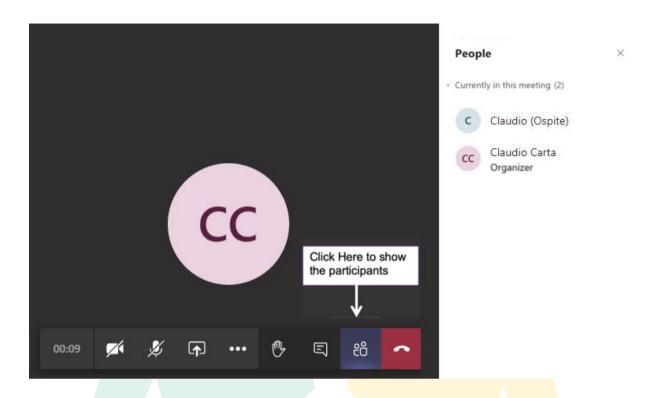
6/ When you click on "Join now" a moderator in the meeting will admit you

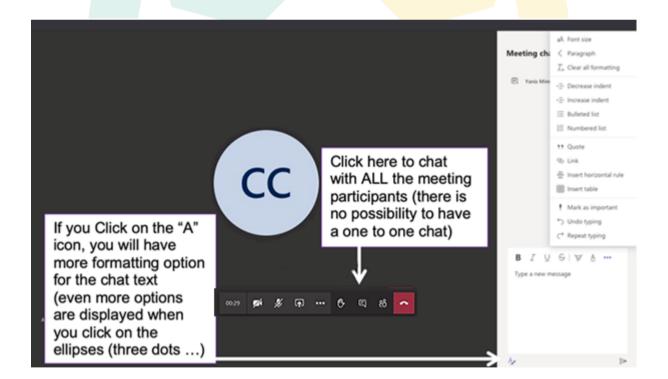


7/ When you join the session you will see up to 4 persons(s) "tiles (thumbnails)" or camera if activated (only one person is present in the below screen capture)







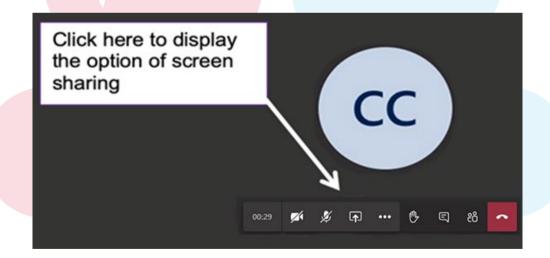




The ellipses (three dots ...) in the bottom bar provide more options. Please consider the option "<u>Turn off incoming video</u>" to improve the quality of your connection (especially if your internet connection is slow):



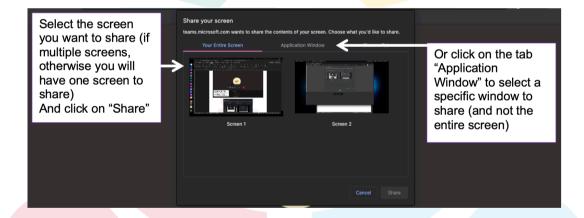
8/ To share your screen:

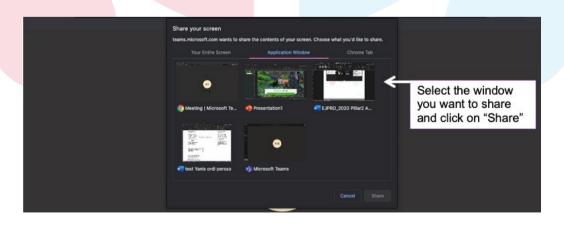




a) On an internet browser:

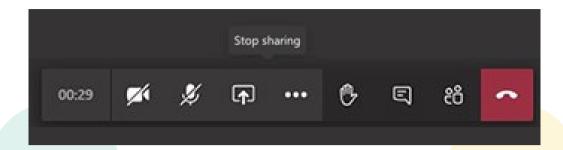






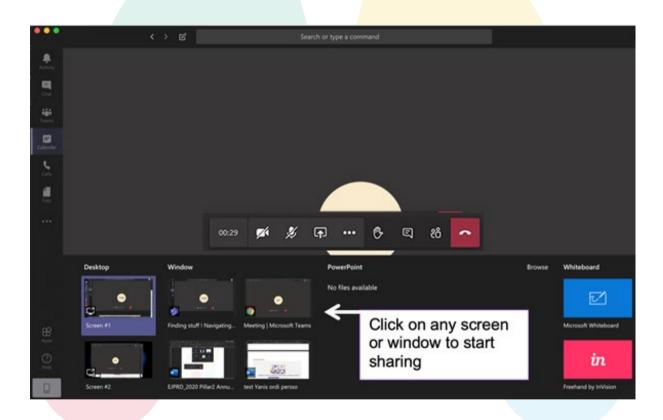


Click on the same icon to stop sharing:



b) On MS Teams desktop application:

You will have all the options displayed at once (sharing screen(s) and the different windows and PowerPoints opened on your desktop)



Please note that with the MS Teams application, when you share your screen, sometimes the application is reduced leaving only a small window that displays the title of the session, your initials and few options at the bottom bar.

Click on that small window to expand the Ms Teams application again