European Reference Network
for rare or low prevalence complex diseases

Network Intellectual Disability and Congenital Malformations (ERN ITHACA)

Manchester Centre for Rare Disease

Jamie aged 11

NHS
Manchester University
NHS Foundation Trust

How virtual health care is happening in ERN-ITHACA

Sofia Douzgou, MD, PhD, FRCP
Consultant in Clinical Genetics and Honorary Senior Lecturer
Tele-Health WP Lead for ERN ITHACA

Connecting with previous experience

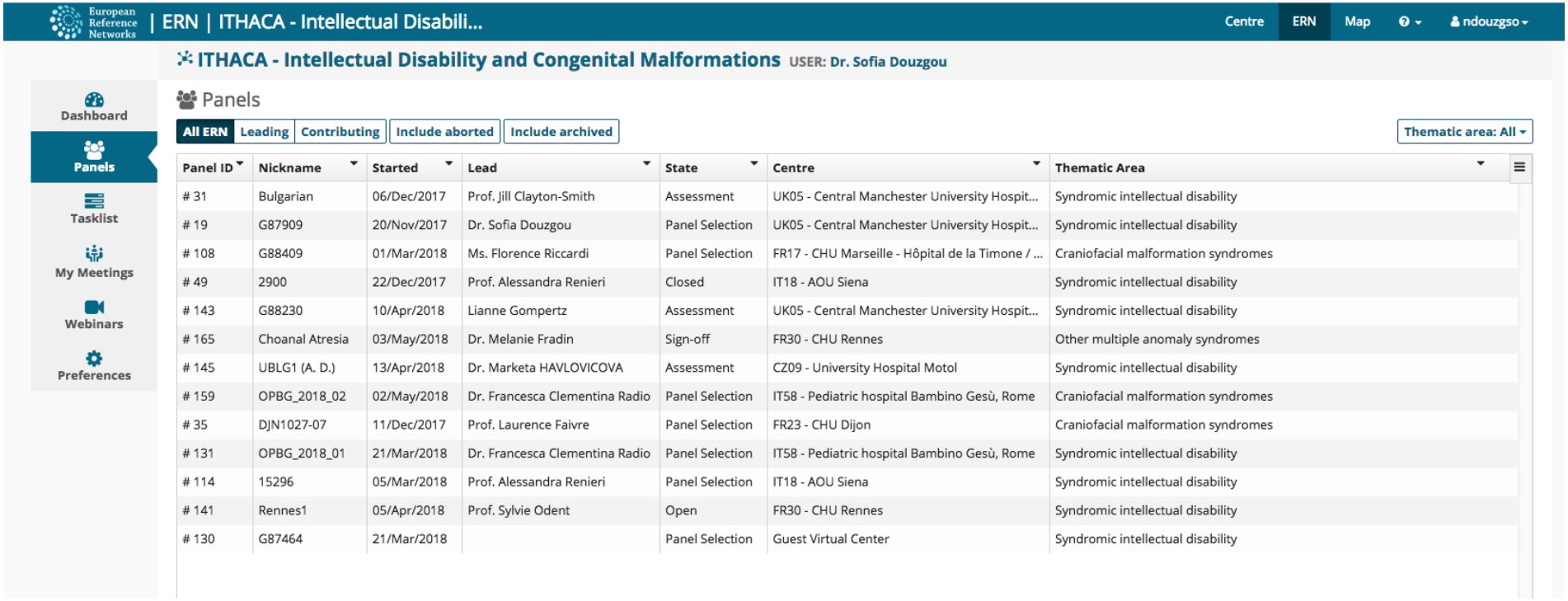
Dysmorphology at a distance: Results of a web-based Diagnostic Service

22.50%	New, Clinical Diagnosis
17%	Recurrence Risk
14%	Diagnosis with available genetic test
5%	Diagnosis of unknown genetic cause
26%	Confirmation of Submitted Clinical Suspicion
17%	Refuted Submitted Clinical Suspicion
1%	New syndrome
70.50%	Genetic Investigations suggested
35.50%	Other laboratory Investigations suggested
26%	Imaging suggested
23.50%	Other specialist opinion suggested
181	Differential Diagnosis offered
1-9/100 000 - unknown	Range of Prevalence of conditions diagnosed
5	Average number of expert reviews
36 days	Average turn-around-time of diagnosis
127 euros	Estimated Cost per case
12	Medical specialties of registered users
39	Participating countries

Douzgou et al, EJHG, 2014

A network of centres of expertise dyscerne for dysmorphology

Harnessing multiple and variable participation



Panel ID	Nickname	Started	Lead	State	Centre	Thematic Area
# 31	Bulgarian	06/Dec/2017	Prof. Jill Clayton-Smith	Assessment	UK05 - Central Manchester University Hospit...	Syndromic intellectual disability
# 19	G87909	20/Nov/2017	Dr. Sofia Douzgou	Panel Selection	UK05 - Central Manchester University Hospit...	Syndromic intellectual disability
# 108	G88409	01/Mar/2018	Ms. Florence Riccardi	Panel Selection	FR17 - CHU Marseille - Hôpital de la Timone / ...	Craniofacial malformation syndromes
# 49	2900	22/Dec/2017	Prof. Alessandra Renieri	Closed	IT18 - AOU Siena	Syndromic intellectual disability
# 143	G88230	10/Apr/2018	Lianne Gompertz	Assessment	UK05 - Central Manchester University Hospit...	Syndromic intellectual disability
# 165	Choanal Atresia	03/May/2018	Dr. Melanie Fradin	Sign-off	FR30 - CHU Rennes	Other multiple anomaly syndromes
# 145	UBLG1 (A. D.)	13/Apr/2018	Dr. Marketa HAVLOVICOVA	Assessment	CZ09 - University Hospital Motol	Syndromic intellectual disability
# 159	OPBG_2018_02	02/May/2018	Dr. Francesca Clementina Radio	Panel Selection	IT58 - Pediatric hospital Bambino Gesù, Rome	Craniofacial malformation syndromes
# 35	DJN1027-07	11/Dec/2017	Prof. Laurence Faivre	Panel Selection	FR23 - CHU Dijon	Craniofacial malformation syndromes
# 131	OPBG_2018_01	21/Mar/2018	Dr. Francesca Clementina Radio	Panel Selection	IT58 - Pediatric hospital Bambino Gesù, Rome	Syndromic intellectual disability
# 114	15296	05/Mar/2018	Prof. Alessandra Renieri	Panel Selection	IT18 - AOU Siena	Syndromic intellectual disability
# 141	Rennes1	05/Apr/2018	Prof. Sylvie Odent	Open	FR30 - CHU Rennes	Syndromic intellectual disability
# 130	G87464	21/Mar/2018		Panel Selection	Guest Virtual Center	Syndromic intellectual disability

- 1st ERN to open a panel with participation from multiple countries
- 13 panels; 2 closed, 11 ongoing assessment
- Panels led by 5 different countries; 1 guest lead from a center that is not a member
- 3 panels led by a trainee; 1 panel led by a trainee who joined the HCP as an ERN bursary
- 4 months of activity: set targets exceeded

Increasing participation rate

	EUROPEAN REFERENCE NETWORK PROJECT STATUS REPORT	Reporting period : MARCH 2018
	CPMS STATUS REPORT	CPMS, version : insight_ern#rel11.01
		System available since: 20/11/2017

CPMS ACTIVITY

NUMBER OF USERS REGISTERED			
This month	63	Total	722

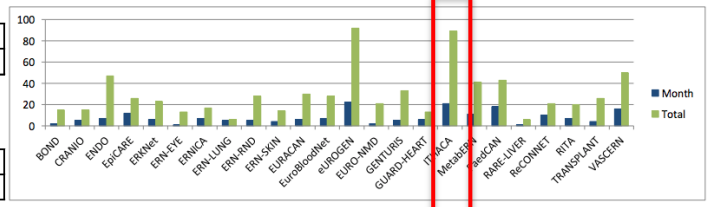
NUMBER OF ACTIVE USERS			
This month	190	Total	717

*Total Active users = Sum of active users/Month

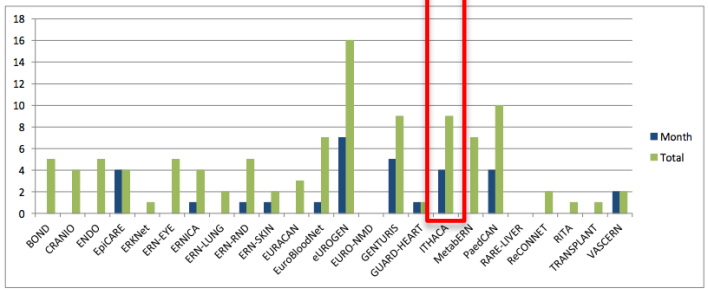
NUMBER OF CREATED PANELS			
This month	31	Total	111

NUMBER OF PANELS CLOSED/ARCHIVED			
This month	6	Total	13

ACTIVE USERS/ ERN



PANELS /ERN



Service usage report for Quarter 1 2018 in relation to CPMS from ERN ITHACA

January 2018: Number of active users*: 16

February 2018: 19

March 2018: 21

*Active users is defined as a count of users that have logged on that month and effected any change to the database or written to the system.

Troubleshooting

- No panels submitted yet by 50% of countries involved in ITHACA (Belgium, Cyprus, Finland, Germany, The Netherlands, Portugal, Romania, Sweden)
- Clinical Lead from the Netherlands refuses to submit cases as they/their HCP do not think that CPMS respects new GDPR criteria
- Most clinical leads involved delegate the submission to trainees who, we have asked, should have their own login credentials
- The 2-factor authentication has been the most difficult aspect for clinical leads to engage with the system! They did not wish to use personal mobile numbers as part of the process.
- Most clinicians have NOT succeeded in completing a standard workflow: most stop/err at the choice of panel and/or request too frequent input from coordinator
- Not ALL activity counts, in fact, multiple forum comments by the same clinician do not count as separate activity.
- We are currently requesting a ‘leaner’ version of cases submission/discussion.

Handling variable queries

- What is the diagnosis?
- Is this the right diagnosis?
- The laboratory result does not confirm the clinical diagnosis: is the diagnosis correct?
- Should we use this type of medication in this patient?
- What is the recurrence risk of the patient's condition for family members?
- Have you seen other patients with this condition?
- The clinical diagnosis has been confirmed in the laboratory but the patient has a symptom that is not part of this condition: is this related?

7/13 active panels with a distinct clinical query: reflection of the diversity of the groups of conditions covered and the rarity of the single disorders

Looking towards the future

- Include training cases
- Filter cases that will benefit from existing research projects
- Choose cases for rare disease registries
- Live tele/e-consultations
- Document relevant usage aspects to pilot health economics

Contact us:

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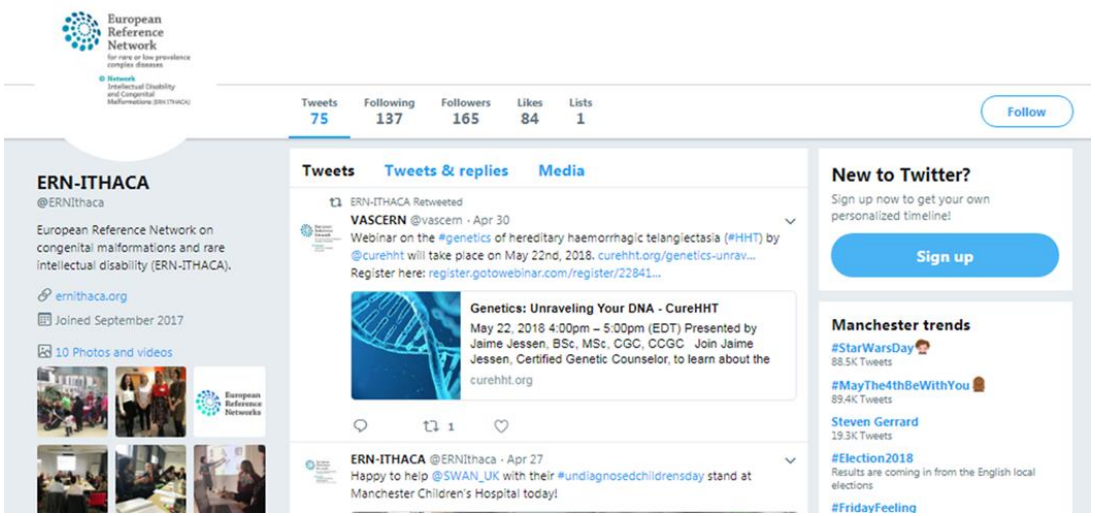

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