ERN-ITHACA: Intellectual Disability TeleHealth And Congenital Anomalies

a European home to end a diagnostic odyssey

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ARTICLE

Dysmorphology at a distance: results of a web-based diagnostic service

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In 2007, the DYSCERNE pilot project funded by the European Commission Public Health Executive Agency (EU DG Sanco) aimed at setting up a network of expertise for patients with rare dysmorphic disorders. As part of DYSCERNE, a Dysmorphology Diagnostic System (DDS) was set up to enable clinicians throughout the EU to submit cases electronically for diagnosis using a secure, web-based interface, hosted at specified access points (Submitting nodes), in 26 different European countries. We report the outcome of this service for 200 cases submitted consecutively between January 2010 and 2012. Each case was reviewed by an average of five expert reviewers. An average of three possible syndromic diagnoses was suggested per case. In 22.5% of the cases, a consensus clinical diagnosis was reached. Genetic testing was suggested in 70.5% of the cases, whereas other laboratory investigations and diagnostic imaging were recommended in 35.5 and 26% of the cases, respectively. Further specialized opinions were suggested in 23.5% of the cases. Overall, a total of 181 very rare or extremely rare genetic syndromes were considered in the differential diagnosis of the 200 cases. In two cases, the reviewers suggested that the findings represented a new syndrome, and in one of these syndromes the underlying genetic cause was subsequently identified. Other benefits of the submission process included the possibility of directing the case submitters to specific centres for diagnostic testing or participation in research and educational benefit derived for both case submitters and reviewers.
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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

The web-based Dysmorphology service was unique in defining new clinical entities which were then confirmed, molecularly.

A network of centres of expertise
dyscerne
for dysmorphology


Helsinki
Stockholm
Vilnius
MCR
Amster
Gronin
Lubeck
Rotter
London
Nijm
Antwerp
Maastricht
Brussels
Lille
Dusseldorf
Leuven
Prague
Paris
Rennes
Tuebingen
Montpell
Dijon
Rom
Padova
Milan
Lyon
Bologna
Bordeaux
Marseille
Floren
Siena
Coimbra
Rome G
Rome BJ
Naples
Troina
Cyprus

BOARD

SECRETARIAT

Affiliate Network

Affiliate Network

Affiliate Network

Affiliate Network

Affiliate Network

Affiliate Network

Affiliate Network

Affiliate Network

Management

Dissemination

Research

Evaluation

Patient and Public Involvement

Management Guidelines

TeleHealth Diagnosis

Teaching and Training

Patient Registries

Milan

London

Bordea

Paris

Rennes

Marseil

Troina

Rotter

Nijm

Masstr

Gronin

Amster

Leiden

Leuven

Rome BJ

Rome G

Naples

Floren

Siena

NoRo

Padov

Monza

Antwerp

Lille

Brussel

Helsinki

Iasi

Stock

Lyon

Tubin

Dussel

Montp

Coimbra

Pecs

Vilnius

Interacting Networks

Interacting Networks

Interacting Networks

Interacting Networks

ADVISORY COMMITEE

The Role Of Our ERN

What is the diagnosis?
What is the best way of investigating and treating this condition?
What health surveillance is needed
Who is doing research in this area?
How can I learn more about it?

What is the cause of our child's problems?
Are we to blame?
Is there a cure?
What does the future hold?
Where can we get the best care?
Is there research going on?

Workpackages

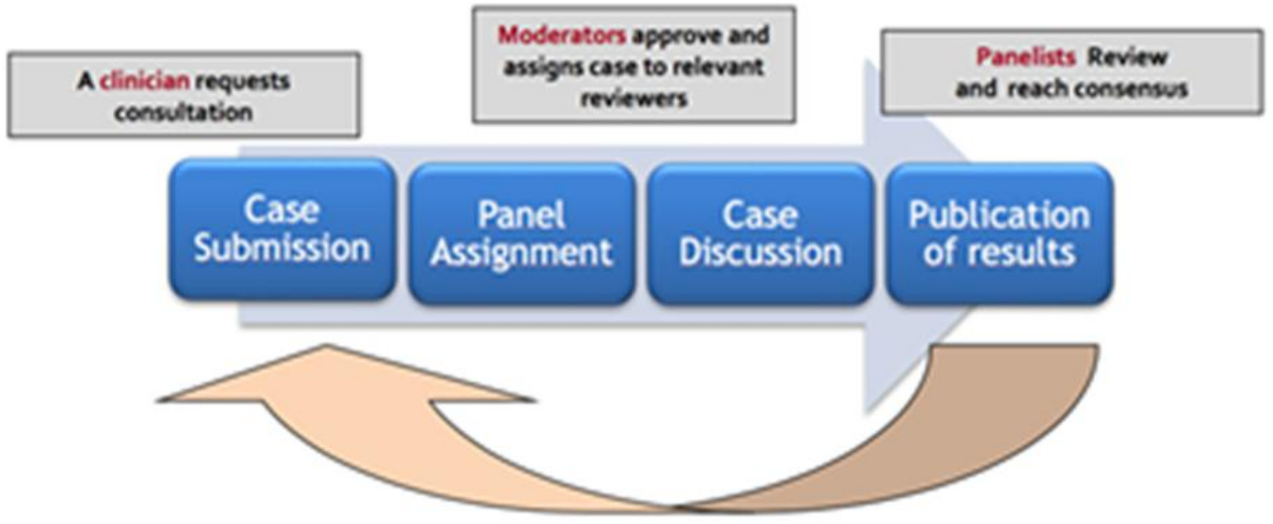
WP1	Coordination of the Network
WP2	Communication and Dissemination
WP3	Evaluation
WP4	Expert Patient Care/Guidelines
WP5	Telehealth
WP6	Patient Registries
WP7	Teaching and Training
WP8	Collaborative Research

A Telemedicine Core: ITHACA

- Electronic submission of clinical cases by clinicians
- Concise history and images/videoclips
- Wide geographical coverage
- Discussion forum
- Facility for live consultations

EXAMPLE OF A POSSIBLE REVIEW WORKFLOW

1. **Case Submission**: online Case submission (image and eCRF)
2. **Panel Assignment:** moderator approval of case and panel assignment
3. **Case discussion:** review by experts (eCRF with structured information and free text), with recommendations synthesis
4. **Publication of results**: case conclusions available for submitting clinician



Positives So Far

Even before the network was approved, parents and professionals have spent many hours of time thinking how they can improve the lives of those with rare disease

We have learned a huge amount from patients about what they want and what they can contribute

We have learned a great deal from each other in gathering examples of best practice

Challenges

Coordinating membership, especially as network increases in size

Setting up a network with no initial resources

Negotiating Information Governance regulations for IT

Multiple languages

Large amount of EU administrative work

Choosing SMART deliverables

Involving Patients

- We have both an E-Pag group and independent patient advisors
- Initial meeting supported by SPARKS
- Patients/parents involved from the start, we incorporated comments into our workplan
- 2 patient/parent members on our Board
- Patients/parents co-leading workpackages
- Re-imbursement of patients

Sparks
THE CHILDREN'S MEDICAL RESEARCH CHARITY

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Thank you !