



WHAT'S NEXT AFTER THE SEARCH FOR A DIAGNOSIS? THE FUTURE OF SPECIALISED HEALTH SERVICES

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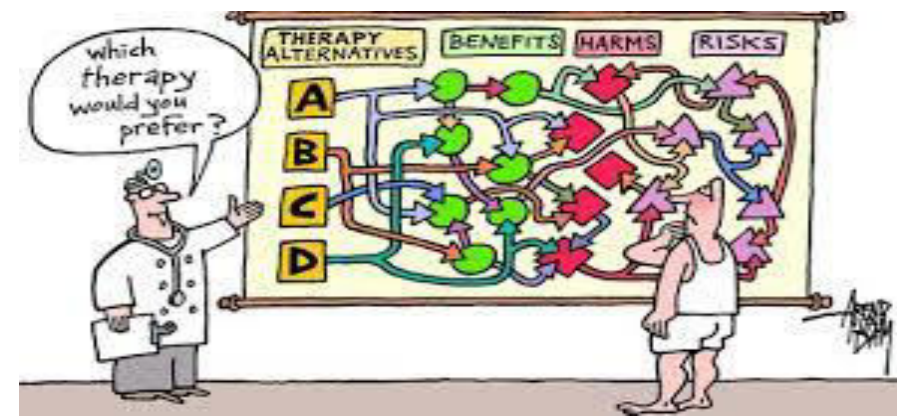
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So, does the patient's odyssey ever end? Ithaca may still be far away.....



- **Finding the expert:** where to seek help, how to navigate health and social care services, geographic constraints, cost of travelling and hours off-work
- **Quality** of healthcare: MDTs, rehabilitation facilities, competent HCPs
- **Education:** comprehension of diagnosis, diversity and heterogeneity of the disease, uncertainty about management and prognosis, chaotic information search on the internet
- **Counseling** by the expert: key parental concerns, realistic expectations, informed decision making and forward planning, family screening
- **Treatment** options: availability of medicines, curable vs incurable disorder, aetiological vs symptomatic treatment, cost, medical aids, lifestyle interventions, participation in clinical trials
- **Psychological** support: how to deal with the disease, impact on family, social stigma, isolation, exclusion, available support and advocacy groups
- **Social** aspects of the disease: schooling, leisure time, future work, relationships, insurance discrimination, few professional opportunities

*Basel D, McCarrier J, PedClin N Am 2017
Rare Disease Facts: EURORDIS, 2005
Litzkendorf S et al, J Rare Dis DiagnTher 2016*



The future of specialized health services

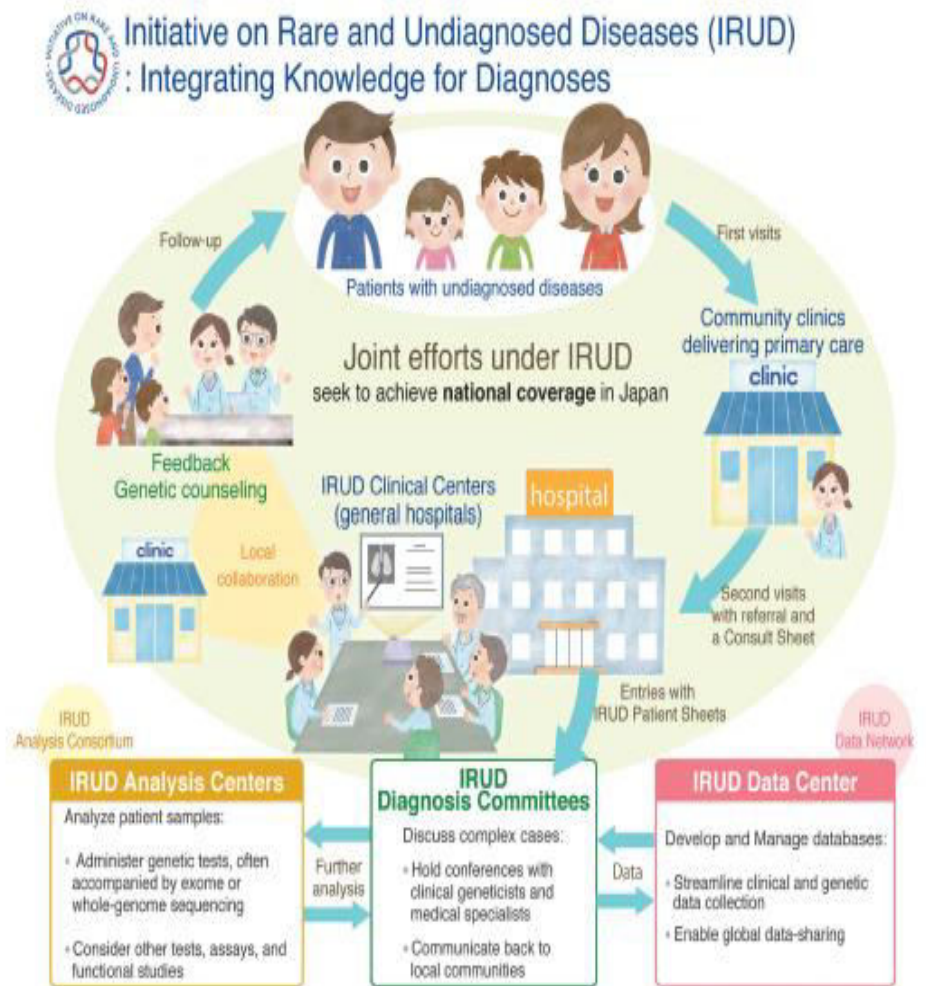
The way forward



- *The Japanese experience (IRUD)*
 - ✓ Data-sharing framework: collaboration between researchers and HCPs
- *The USA experience*
 - ✓ Establishment of non-profit, foundation-sponsored clinic networks to facilitate access to specialized care
 - ✓ Evaluation criteria: clinical expertise, availability of MDTs, patient volume, involvement in research
 - ✓ The NF1 clinic paradigm: in 10 years, 56% increase of certified NF clinics and 51% increase of NF1 patients treated, better clinical outcomes, more funding, smooth transition to adulthood
- *The European experience (ERNs)*
 - ✓ “Connecting the dots” between countries, experts and patients with virtual networks, aiming to tackle rare diseases that require highly specialized treatment and a concentration of knowledge and resources
 - ✓ Platform for the development of guidelines, training and knowledge sharing
 - ✓ Access to >900 expert centers, 24 thematic networks and virtual advisory boards
 - ✓ Mobility of expertise across borders; the patient need not travel to find the expert
 - ✓ The European Commission supports member states by pooling knowledge and expertise, registries, data and funding

The vision

Evidence-based, patient and family-centered care through all stages of life



Final remarks: key strategies for the future

- Encourage patient voices to be heard, organize psychological workshops and distribute educational materials
- Endorse national and international collaborations, organize clinical decision support groups, appoint liaison officer for rare diseases to ensure better follow up care
- Engage in pioneering research
- Use digital data and the cutting-edge technology (IT systems)
- Adopt a wider policy alignment, to include mental health support and social care

Adachi T et al, Eur J Hum Genet 2017
Merker VL et al, BMC Health Service Res 2018
ERN brochure, European Commission 2017
Reuter CM et al, J Pediatr 2018

