EQA for Rare Diseases

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RARE DISEASES – facts and figures

- prevalence <5/10,000 people in Europe
- ~300 million people worldwide
- >6,000 rare diseases known today
- extremely heterogeneous:
  72% genetic, the remainder are rare tumors, immunologic and infectious diseases, as well as rare malformations, among others

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RARE DISEASES – general challenges

– resources and expertise are as scarce as patients

– very long average latency before the right diagnosis is found (if at all)

– no specific treatment for the vast majority of diseases >> **finding a diagnosis is still extremely relevant!!** (psychologically, access to social services, clinical trials / research…)

Challenges in laboratory medicine

- low throughput (wide range)

- development of automated testing systems /kits not lucrative for companies (in-house tests used widely)

- labor-intense, time-consuming, often manual analyses >> high costs, variable quality

- timely reporting is a critical factor

- interpretation of results by lab expert usually required (taking into account clinical data, pre-analytics, method applied) >> close collaboration between lab expert and clinician necessary

- not normally part of routine training in laboratory medicine

- clinicians often not aware of these issues

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Current situation

- many small labs performing these analyses still not accredited

- special demands for rare disease diagostics typically not included in accreditation norms (minimum number of analyses performed annually, competence assessment of personnel)

>> rare diseases are an exceptional challenge for quality assessment!!

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Challenges and demands for EQA in rare disease diagnostics

- scarcity of sample material (patients are rare, children,…; at the same time often large quantities needed for manual testing)
- less-than-ideal sample condition due to non-standardized pre-analytic processing (age of sample, repeated freeze-thaw, long transport,…)
- no EQA available for many tests/diseases even as international schemes

- frequent participation in EQA desirable (e. g. patient samples tested only against selected positive control for extended period of time, critical analyses like pre-natal/pre-implantation testing, no standardized methods,…) but typically not possible (not available, costly)

- variability (test systems, reference ranges) should be taken into account in evaluation of EQA results

- assessment of pre- and post-analytical steps, timely reporting

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>> huge wish list, many to-do's

Can be solved only by targeted policies and specific designations for laboratories performing analyses for rare diseases, in combination with EQA

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