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Mainstreaming Genetics

The UK story

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House of Lords Genomic Medicine Report 2009

"Genomic medicine will clearly have a huge impact on health provision and the NHS in particular over the next few years"

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What do we mean by mainstreaming genetics?

- Strategy for the most effective use of genomic knowledge and technologies to benefit patients in different clinical specialties.
- Incorporation of genetics into the standard practices of mainstream medicine.
- Likely to involve some sub-specialisation for inherited conditions
- Changes to existing pathways for some acquired conditions
- Genetics services are and will remain an essential element of this paradigm

- Already there are areas of ‘good practice’ including joint clinics in which specialties work side by side with genetic services to provide care for patients with inherited disorders

- National projects: DDD &EACH, 100,000 genomes

- Haematological malignancy where genetic testing is already embedded in a number of care pathways BUT…….

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Considerations

- Areas of recognised clinical necessity not necessarily supported by robust and future-proofed funding

- Savings in one area of the patient pathway not matched by supporting investment in the evolving test area

- How can we facilitate the translation of these advances into clinical practice, including the operational changes needed to bring genetic testing into the mainstream?

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Drivers

- The Mainstreaming agenda is being driven centrally by the DoH through the arrangements of the NHS CB.
- Mainstreaming transfers the responsibility for diagnostic testing, including the responsibility for financing, to the specialities other than Clinical Genetics.
- The development and implementation of Next Generation Sequencing will have a profound impact on the delivery of diagnostics throughout medicine and is likely to affect every speciality, either directly or indirectly.

Commissioning Implications

- Clinical Genetics activity and all associated genetic testing fully funded through the NHSCB
- Clinical Genetics will remain responsible for extended family testing, including predictive pre-symptomatic testing, syndrome diagnosis involving multiple organ systems or complex dysmorphology, prenatal diagnosis and inherited cancer.
- ALL other diagnostic testing to be requested and funded by referring speciality
- However the emphasis will be very much on a MDT model with Clinical Genetics supporting and advising where required across the specialities.

- Introduction and exploitation of new technologies is dependent on a fully functioning commissioning process.
- Need to be able to facilitate the translation and integration of new diagnostic methods
- Further develop multi-disciplinary collaboration
- Public health physicians are used to population-wide measures such as screening

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Transitional issues

- All activity from external Trusts will be invoiced
- Referring Trusts will not receive additional funding so will need to manage demand and/or seek alternative cheaper providers.
- May be a shortfall due to a differential between the contract value and the activity value at the prices currently charged.

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New technological challenges

- NGS poses a significant challenge in the management of the data, both practically in terms of its quantity, and prognostically and diagnostically in its interpretation.

- Ethical considerations of UVs – patients right to know or not to know

- Consent – when and what for

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Other Challenges

- Supporting the integration of genetic services
- 60% people are likely to develop a disease, which is at least in part genetically determined. Hayden EC (2012)
- Changing demographics: ageing population, increasing number of cancer cases
- Ambiguity over service provision for acquired disorders
- NGEDC – DH funding for 2 years from 2010 – raise awareness, provide information and tools, develop workforce competencies – but project too short to realise the full potential of this initiative

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Further challenges

- Evidence to support the need for new/integrated services
- Delivery of more genetic testing outside Clinical genetics
- Organisational change –takes time!
- Training implications

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Costs

- Genetic tests relatively expensive – particularly if the gene/mutation is unknown
- Cheaper to test for a known familial mutation
- Tariffs for some conditions should include the costs of genetic testing BUT specifics of this not clear
- May result in increased number of tests requests without the funding to cover it

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BUT important to consider

- Does the result confirm a diagnosis that changes treatment
- Potentially allow identification of other ‘at risk’ family members
- Potentially stop other, more invasive tests
- Potentially curtail a diagnostic odyssey
- Provide sensitive monitoring
- Guide targeted treatment regimes

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Key points

- There will be a significant amount of work to clarify both the diagnostic and funding pathways.
- Key will be to engage with requesting clinicians and their Trusts to ensure that they are aware of the funding changes and the commissioning in place to meet cost of testing.
- Development of Bioinformatics resource as an integral part of the diagnostic pathway
- **Cross specialism collaboration vital**

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Reference documents

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- Future Trends Overview (2012)– The King's Fund
- NICE guidelines: Familial Breast Cancer (update) Draft 2013
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