

New Perspectives in DNA service

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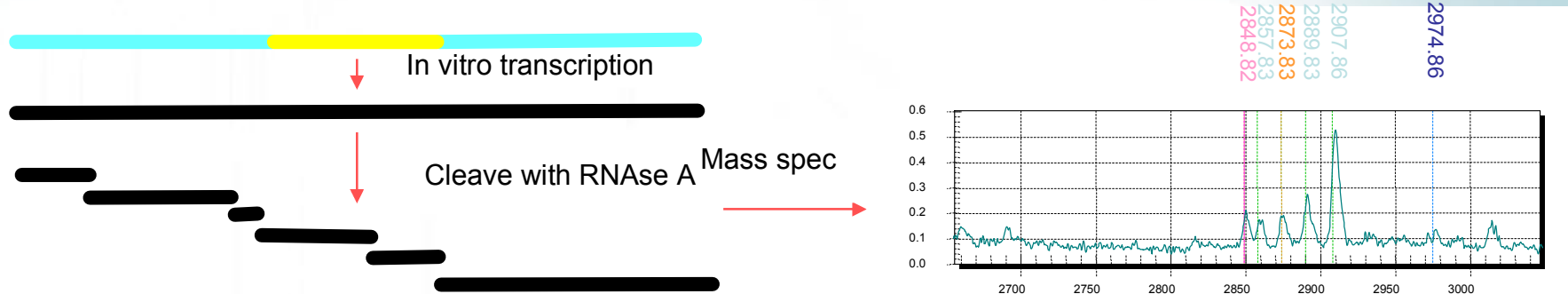
*Setting standards
in analytical science*

- LGC – long history of analytical science – pleased to sponsor the first InSiGHT Conference
- Strategic partnership with Newcastle – add value in medical genetics
- Sequence screening for mutations in large, multi-exon genes
- Transition from research discovery to routine, quality assured, diagnostic service.
- Reduce cost and reporting time, maintaining accuracy – new technical approach
- Starting point the HNPCC genes – MLH1, MSH2, MSH6

Mass spec SNP discovery by comparative analysis of microsequences



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Computer

ACTGCAGCTTCAGATAGTCAGACCCAAAGT

ACT GCAGCT
T CAGAT
AGT CAGACCCAAAGT

Measures masses

↓
Compares masses → LGC FindSNP software suite + Sequenom Discovery Software **identify and assign mutations and SNPs variants**

↑
Calculates masses →

Mutations generate new fragments in up to four cleavage spectra



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5' - ...GATTGAGAAC [T/C] GGTACGG... -3'
3' - ...CTAACTCTTG [A/G] CCATGCC... -5'

T (forward)

T (forward) wt: G2T1 1015.6 Da
5' - ...GATTGAGAAC**T**GGTACGG... -3'

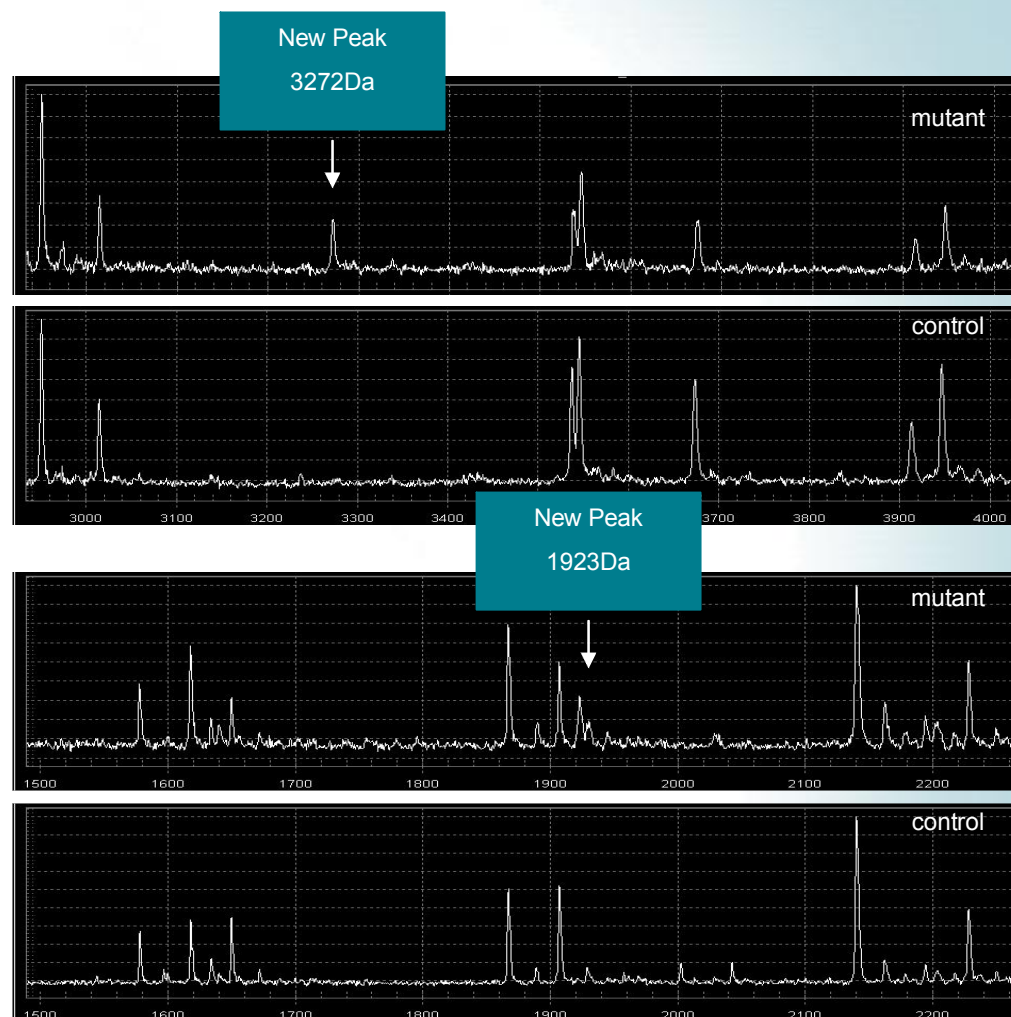
T (forward) wt: A3C1G2T1 2292.4 Da
5' - ...GATTGAGAAC**T**GGTACGG... -3'

T (forward) mut: A3C2G4T1 3272.0 Da
5' - ...GATTGAGAAC**C**GGTACGG... -3'

T (reverse)

T (reverse) wt: A2C2G1T1 1907.2 Da
3' - ...CTAACTCT**T**GACCATGCC... -5'

T (reverse) mut: A1C2G2T1 1923.2 Da
3' - ...CTAACTCT**T**GCACCATGCC... -5'



LGC service



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- Blind studies on MLH1 - collaboration from Newcastle, Leiden and London
- Validated service now available – more info at the stand
- MSH2, MSH6 completing validation shortly
- Nomenclature converter available shortly on web site

- Other services - interest to learn of other gene tasks we could develop a service for

