

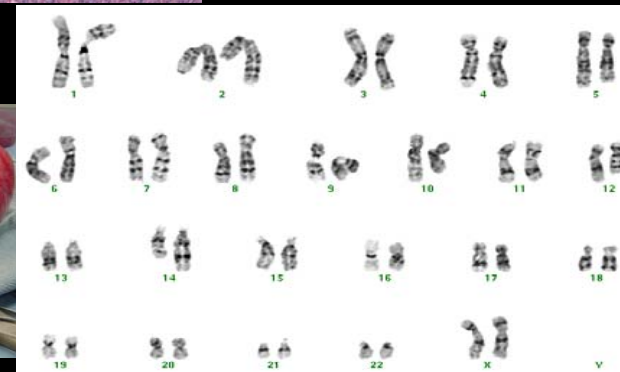
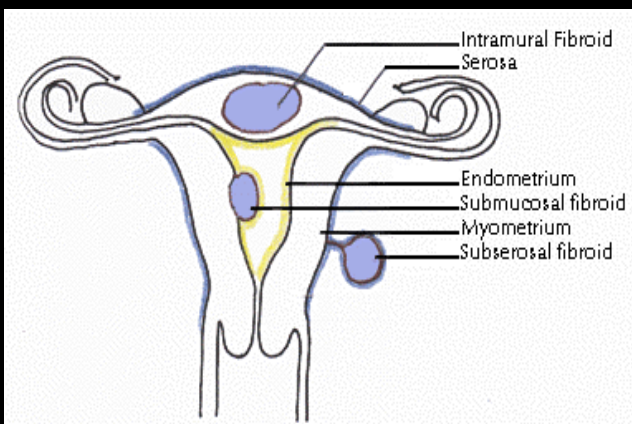
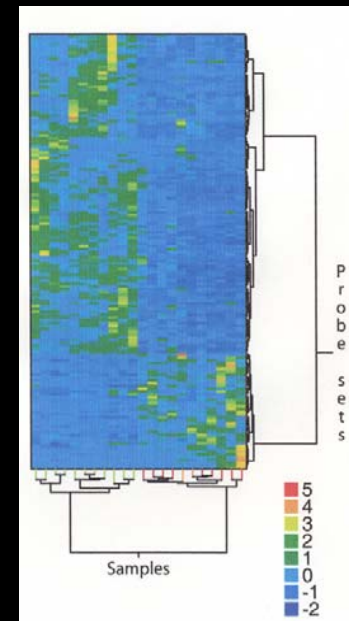
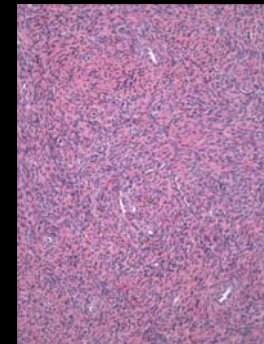
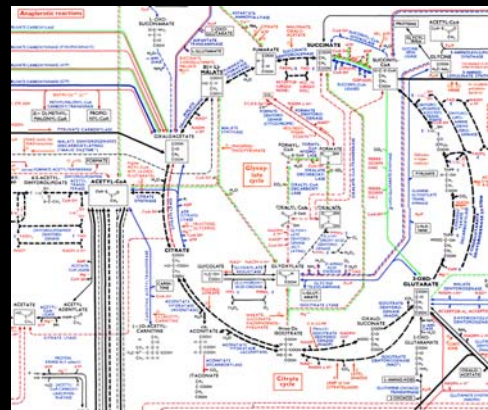
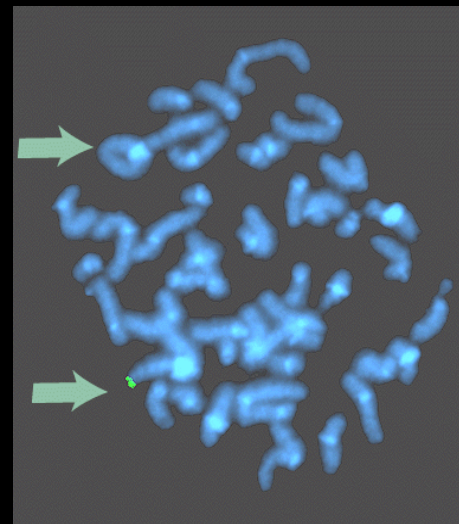


Genetic Links

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How does the *Genotype* affect the *Phenotype*?

- Tumor genotype
- Constitutional genotype



Cytogenetics of Uterine Leiomyomata

- 60% are 46,XX
- 40% are chromosomally abnormal:
 - trisomy 12
 - t(12;14)(q14-q15;q23-q24)
 - del(7)(q22q32)
 - rearrangements of 6p21, 10q22, 13q21-q22
 - deletions of 3q and 1p



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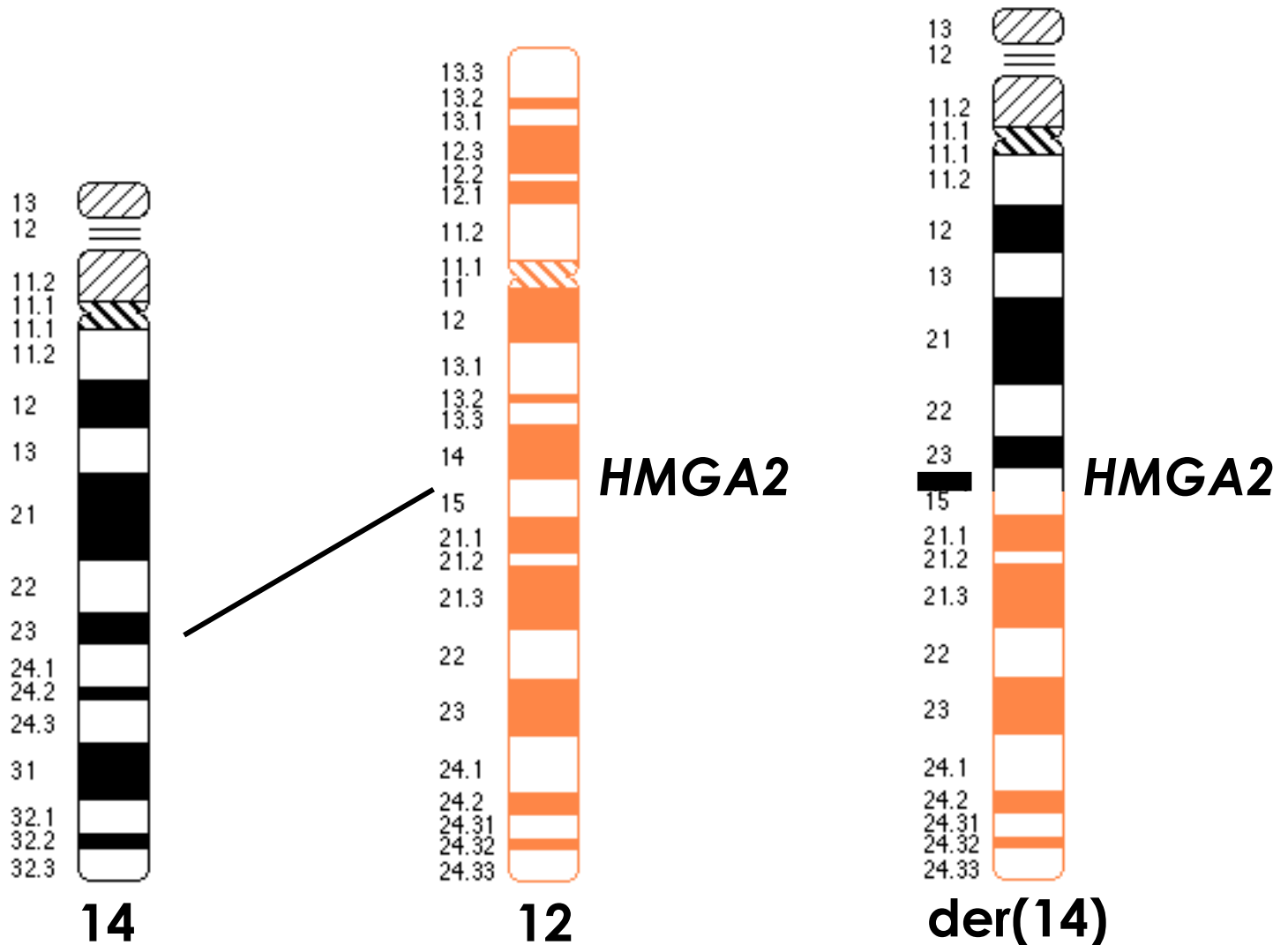
22



X

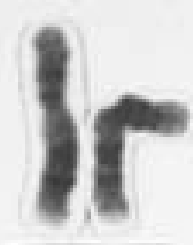
Y

Idiograms of t(12;14) and *HMGA2* localization





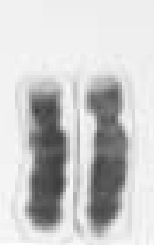
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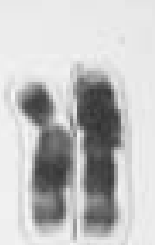
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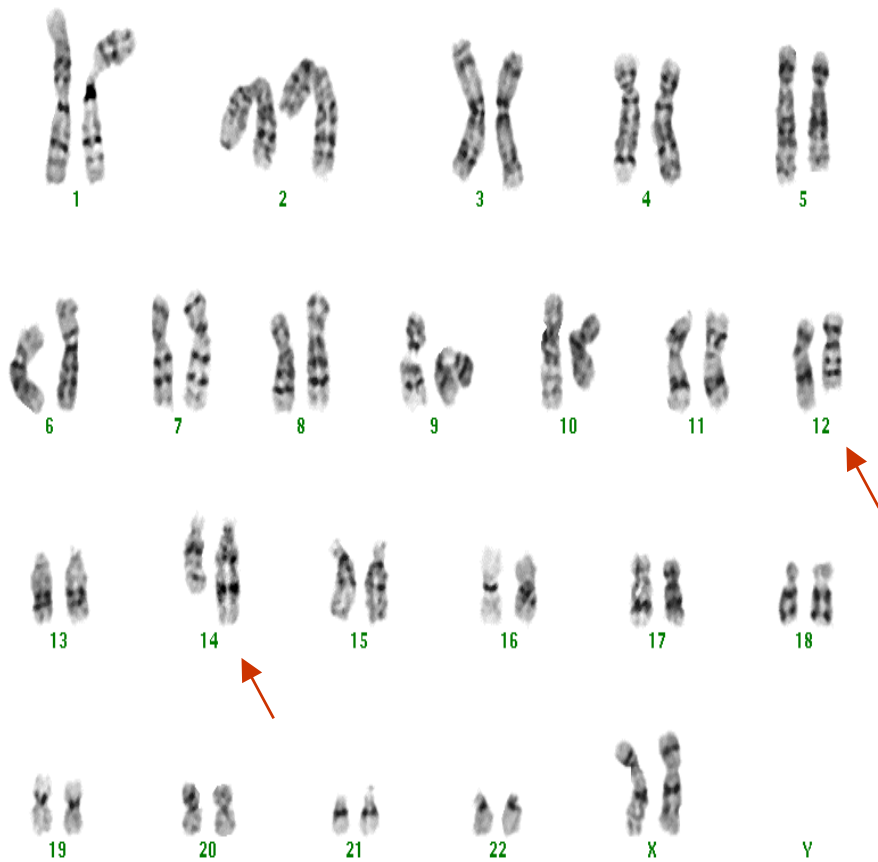


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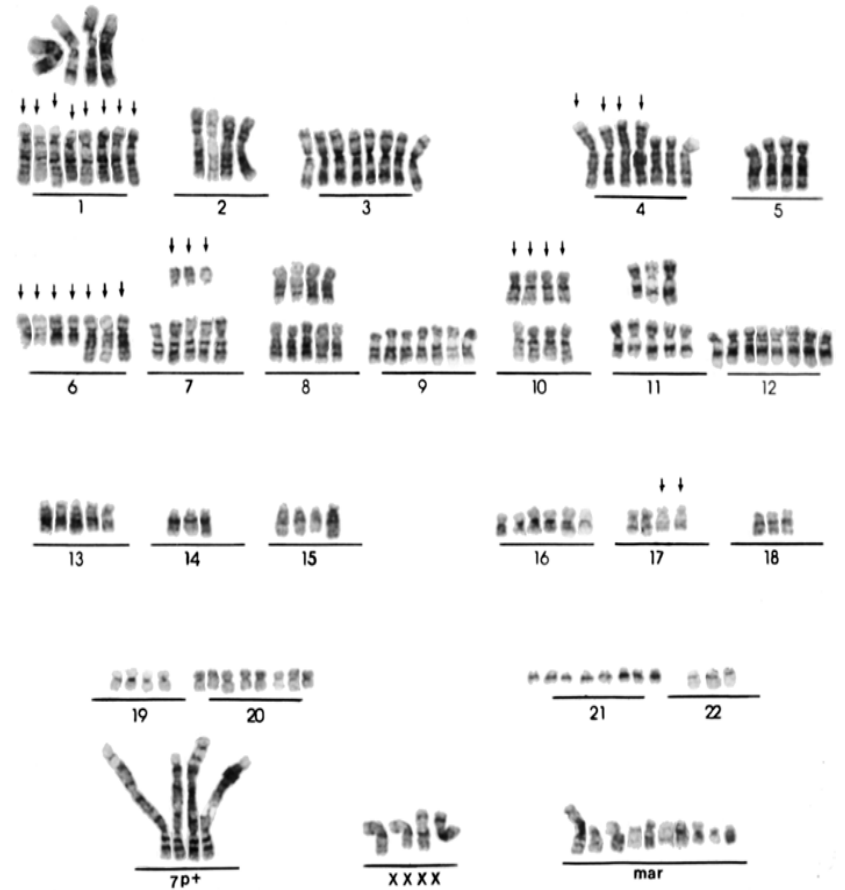


XX

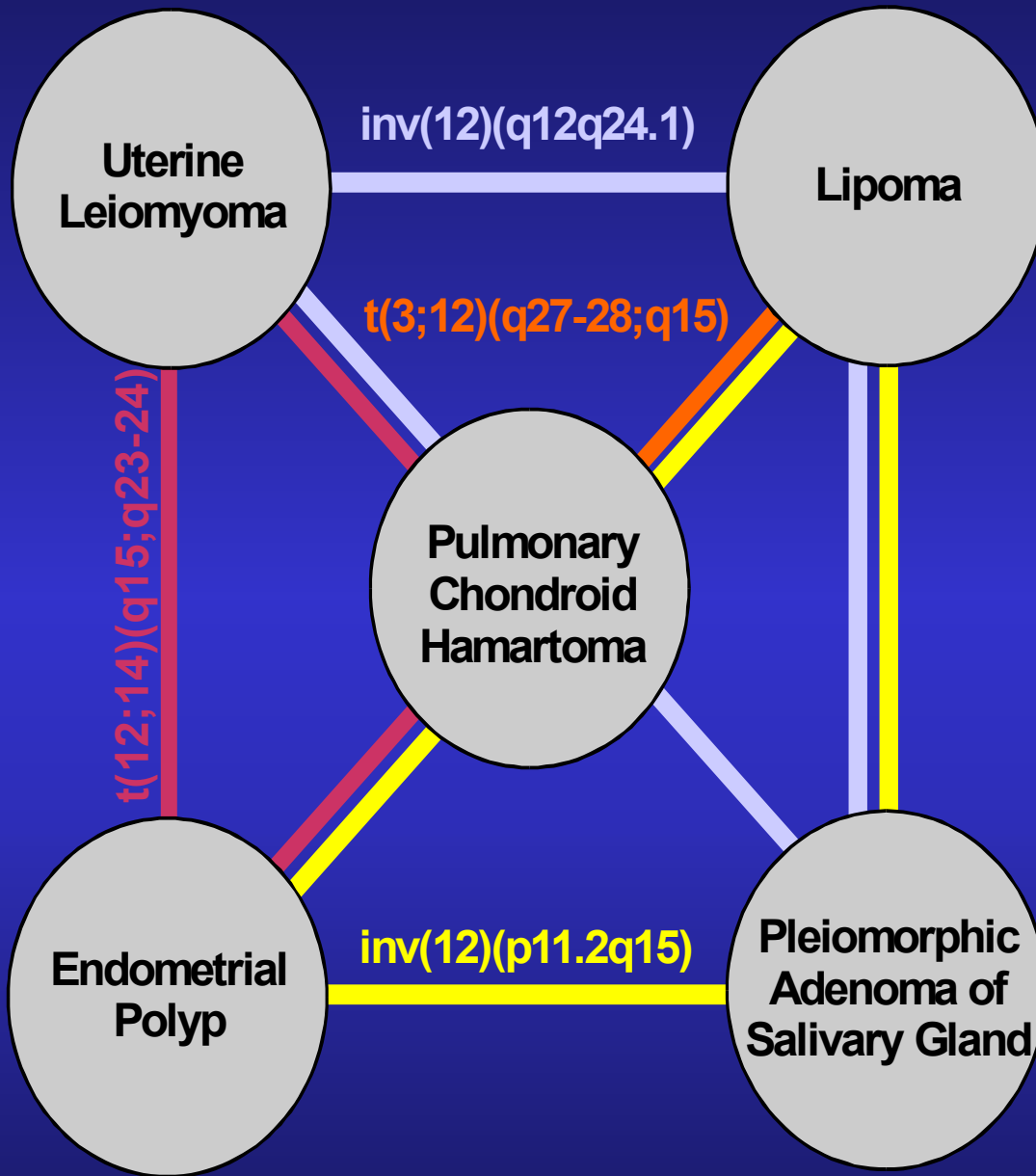
Cytogenetic Analysis of Uterine Smooth Muscle Tumors



Leiomyoma



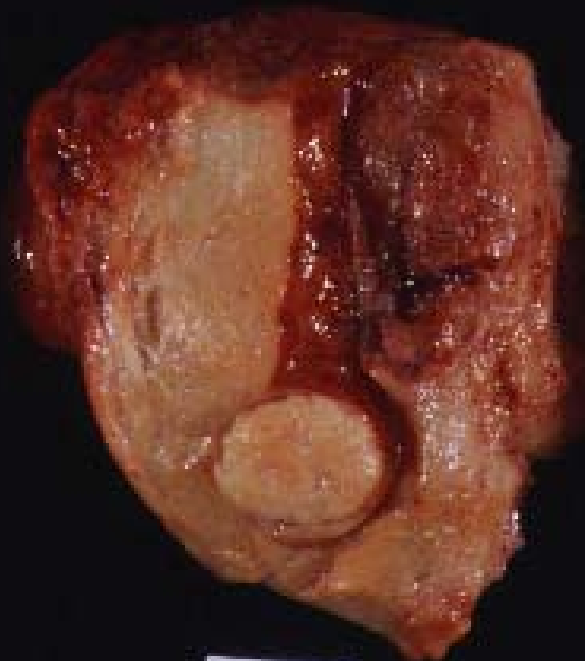
Leiomyosarcoma



0 1 2 cm 3 4 5
BWH Pathology
G95-33589



ST95-199



ST95-202



ST95-200



ST95-201



ST95-203



ST95-204



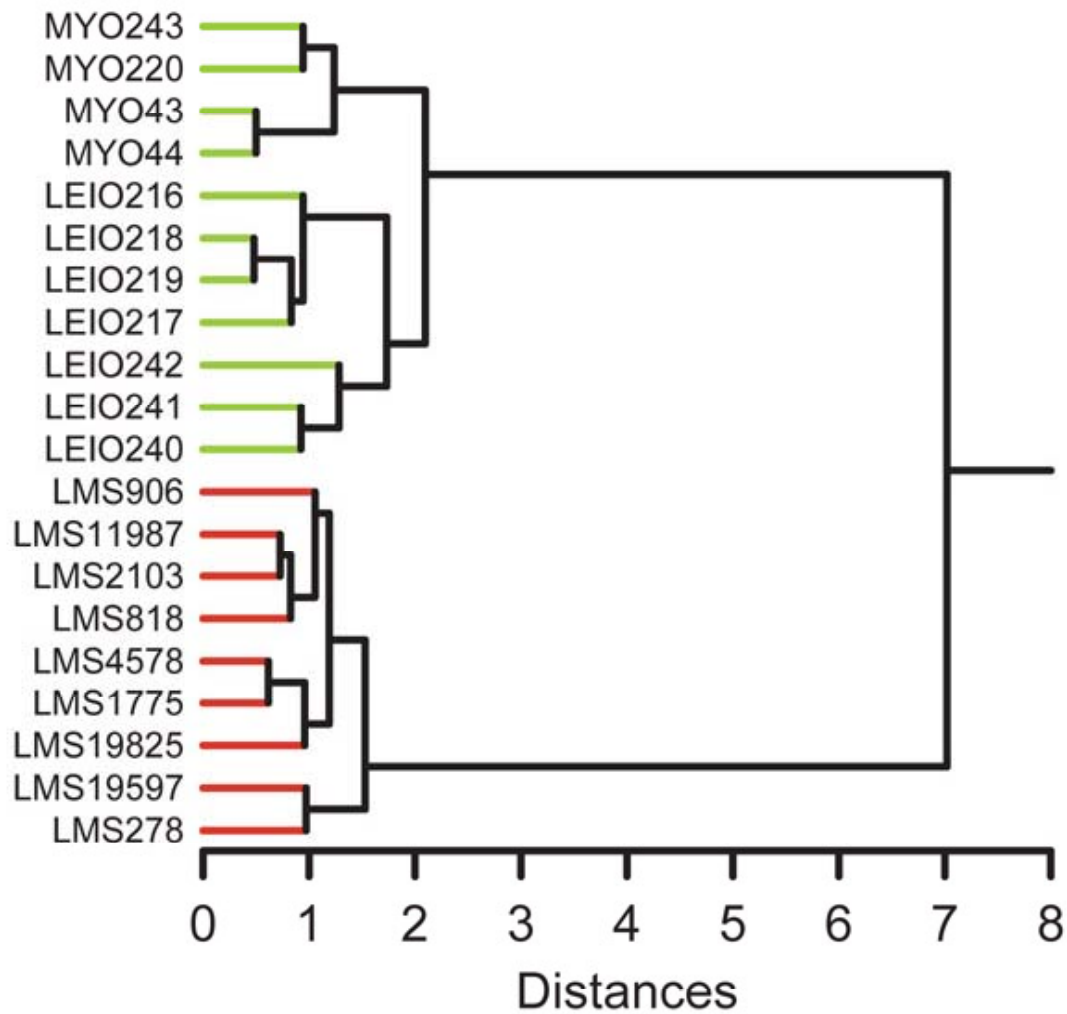
ST95-205

Summary of Myoma Size and Cytogenetic Analysis

| Cytogenetic Analysis | Myoma Diameter (cm) Mean + S.D. | Myoma Diameter (cm) Median | Calculated Myoma Volume (cm³) Mean + S.D. |
|-----------------------------|--|-----------------------------------|---|
| Normal (n=73)* | 5.9 ± 4.2 | 5.4 | 334 ± 1123 |
| Abnormal (n=41) | 7.6 ± 5.3 | 7.0 | 642 ± 1638 |
| Mosaic (n=21) | 5.0 ± 3.0 | 4.5 | 136 ± 165 |
| Non-mosaic (n=20)* | 10.2 ± 5.9 | 10.0 | 1173 ± 2246 |

*p<0.01

A



B

