Familial Medullary Thyroid Cancer (FMTC)

Where did it come from?

In 1968, at the age of 67, <u>Clara Pearl Astle Carling</u> was diagnosed with Medullary Thyroid Carcinoma. With the work <u>Dr. Devaprabu Abraham</u> at the University of Utah, it was later found that this uncommon form of thyroid cancer was caused from a genetic variant. Over the course of many years, several of her children developed the same type of thyroid cancer.

Fast forwarding to recent years, several of <u>Grace Emily Astle Frank's</u> children were also diagnosed with the same Medullary Thyroid Cancer. Since Pearl and Grace are sisters, it became readily apparent that this gene was passed to them from either their father, <u>John Francis Astle</u>, or their mother, <u>Lauretta</u> Hepworth Astle.

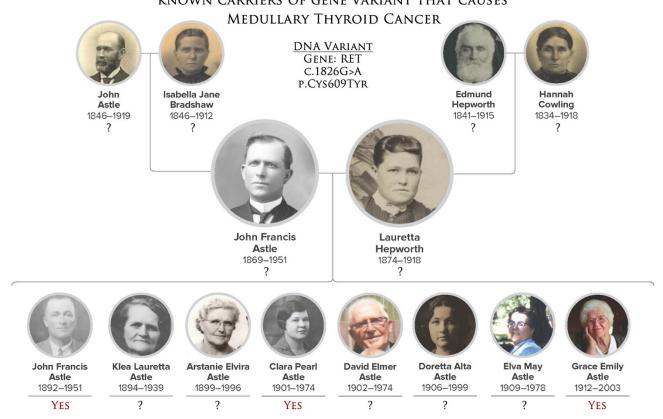
John and Lauretta Astle had a total of thirteen children. Of those children, only eight went on to have children of their own. At this point in time, we know that three of these eight children were affected by this genetic variant (<u>John Francis Astle Jr.</u>, Clara Pearl Astle Carling, and Grace Emily Astle Frank).

While there is much to be learned, we do know that nearly twenty immediate descendants of Clara and Grace have been diagnosed with this same cancer. Many others who have been found to carry this same genetic variant have had their thyroids removed to avoid development of this cancer. This genetic variant is also associated with hyperparathyroidism (identified in the John Francis Astle Jr line), pheochromocytoma, and other endocrine disorders.

If you have knowledge of thyroid cancer or parathyroid disease occurring in the descendants of any of the following people, please <u>contact us</u> to help further the research on Familial Medullary Thyroid Cancer.

- John Astle
- Isabella Jane Bradshaw Astle
- Edmund Hepworth
- Hannah Schofield Cowling Hepworth
- Klea Lauretta Astle Baer
- Arstanie Elvira Astle Nye
- David Elmer Astle
- Doretta Alta Astle Hoskin
- Elva May Astle Kendrick

Family Lines: <u>Astle - Hepworth - Bradshaw - Cowling</u> known carriers of gene variant that causes



DNA Variant

Classification: Pathogenic

Gene: RET (REarranged during Transfection) Nucleic Acid Change: c.1826G>A; Heterozygous

Amino Acid Alteration: p.Cys609Tyr

www.AstleHistory.com

www.HepworthHistory.com