Another high-profile paper on FSHD was released on-line on April 26, 2012, at the journal Cell. Cabianca et al., A Long ncRNA Links Copy Number Variation to a Polycomb/Trithorax Epigenetic Switch in FSHD Muscular Dystrophy, Cell (2012), doi:10.1016/j.cell.2012.03.035. The print version well be out on May 11.

The paper comes from Drs. Davide Gabellini and Daphne Cabianca in Milan, Italy. This is excellent progress made as the result of all of our combined efforts and the efforts of Davide and Daphne. Drs. Gabellini and Cabianca started their work on FSHD as a post-doctoral and pre-doctoral fellow respectively. Davide started as a FSH Society Marjorie and Gerald Bronfman fellow and Daphne started under a FSH Society Festive Evening of Song fellowship. The Society is acknowledged in the paper.

This paper can be found at http://www.cell.com/newarticles and http://www.cell.com/fulltext/S0092-8674%2812%2900463-1.

The current paper from the Gabellini lab is promising for a number of reasons. First, it provides a potential and important therapeutic target, DBE-T, that could potentially be modulated to control disease. Second, it provides a unifying model to explain a number of previously discordant observations, built on two decades of experiments following the discovery of the D4Z4 deletion.

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Cabianca et al., A Long ncRNA Links Copy Number Variation to a Polycomb/Trithorax Epigenetic Switch in FSHD Muscular Dystrophy, Cell (2012), doi:10.1016/j.cell.2012.03.035

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