

## Abstracts of Recent Research

- a. Already for a long time the role of inflammation in the development of FSHD has been discussed, but hard evidence was not provided, mainly because to technical shortcomings and the heterogeneity of FSHD. Now the known group of Ricci provided evidence using microdialysis. The main result is the upregulation of chemokine CXCL13. In principle this opens the way to test whether down regulation of this chemokine will slow down disease development.
- b. At the previous meetings of Dutch FSHD foundation, Vissing [Denmark] and Heerschap [Holland] demonstrated that MRI [Magnetic Resonance Imaging] may become a good method to monitor disease progression and [hopefully soon] the benefit of one of the various routes tested in [pre]clinical trials. Now Vissing's group published a nice article based on the 1-year follow-up of 45 patients. This study demonstrated that MRI is even better than the often used muscle strength and endurance tests.
- c. Various times it has been observed that there is a difference between disease progression in men and women [progression in men is faster]. Now a combined study of Italian and French scientists may have found out why this is the case. Estrogen, a steroid hormone is able to reduce the translocation of DUX4 to the nucleus. It has been shown before that translocation to the nucleus is necessary for DUX4 induced dystrophy. Although this was work with myoblast, it may be important to follow these studies, as existing medication is available to change hormone balances.
- d. The former group of Alexandra Belayew -one of the scientists identified DUX4 as a major factor in FSHD, published a good paper on the potential of anti sense oligo's against DUX4. In principle anti sense oligo's can bind to RNA and then block the translation of the RNA into protein. So anti sense oligo's against mRNA encoding DUX4 will prevent DUX4 protein formation. Previously nice results have been published by the group of Harper Ohio USA, but this paper shows really the potential of this; however the problem remains to deliver these molecules at the right time at the right place. Fortunately solutions for this are in progress.

What I like to know whether this is about what you expected, whether the official summaries [added as scans] and my summary are useful to you as patient. If you have comments please send me them by mail, if you like to have the full paper, I can send that to you.