EDAR Variant—Living in Malta

(By Stephen Mifsud)

I have written several articles about the flora of Malta, being a professional Maltese Botanist qualified by a Masters in Biodiversity and taxonomy of plants from the University of Edinburgh, but I would have never imagined myself writing an article about myself and Ectodermal Dysplasia. One reason being that despite in my early 40s, I've discovered about this condition only a few years after the birth of my cute 9-year old daughter, when I noticed that she had the exact pattern of missing teeth that I and my mother have. I hoped that it might be just a coincidence, but when my second child, now a smart 6-year old boy, had again the same pattern of missing teeth, I was seriously concerned and I have been writing more regularly to the ED group in the UK. One of the many suggestions I received from this excellent support organisation was to perform a genetic test to confirm ED and to which trait it belongs.

First, I performed the test on myself in 2013 and then on my children in 2014. ED was confirmed in all three of us. For me that was no news, just a hope-killer, because from the symptoms I was sure it must have been ED. However, the results still had a piece of news for me because the ED we carry seems to have never been reported, and whether undocumented or new to science, we still have to see. For scientific purposes, the result of our ED is EDAR c. 1264 G>T variant, dominant – just a single base substitution, as rare as winning the national lottery three times in a row!

This ED is very mild and almost without any symptoms, from birth to the start of teenage years. In all three of us (and assumingly my mother) the main visible symptom during childhood is lack of all four lower incisors and only two large incisors above. Teeth are evenly spaced and quite cute, there are no signs of missing teeth when smiling or even laughing, and eating without trouble. Nails, hair, sports activity and social life was perfect.

Unfortunately, I am seeing a developing symptom in my daughter which was neither manifested in me or is visible in my son, and I cannot tell if my mum had it, so maybe it effects females more. My daughter has obvious signs of late development and it was symptomatic mostly at school, when she could not cope with the same pace of her peer pupils. Reading was the worst. Thinking about it, she was late in many other tasks such as urine control, speech, vocabulary, reading (still a major problem at her age) and problem solving. Myself, I never liked reading, and at this present day I can't read more than five pages in one go. Her first milk tooth was replaced at the age of eight and a half years (normally six to seven). However, she is very astute, super-man vision, and difficult to beat her in memory card games (match a pair). She has a visual intelligence much above average, and she can remember such small details of experiences that happened many years ago. These are all good characters for a botanist!

So it seems for me that she has a slow biological clock, and her biological year does not consist of 12 but of 16 months. This is likely to have a negative effect for her progress in school and college, and she has already done a repeat in year 2. My son does not seem to show evidence of this and I was amongst the smartest in class.

In my case, a wave of secondary symptoms started after puberty, taking place very gradually but steadily. The first to appear was the falling of hair, and this was without doubt the most drastic of them all. I had beautiful, although uncared for, hair that was strong, plentiful, wavy brown and fast growing through all of my childhood, normal for Mediterranean boys. From fourteen it started to fall, and I remember very well that I used to have lots of head itching whilst doing my homework, and I would pick up a bunch of hair from the desk and toss it in the toilet with despair.

My self-esteem was decreasing rapidly and I was super aware of my looks. On the other hand, facial hair (beard) was normal. I had scanty normal body hair only on my thighs, pubic area and slightly in the armpits, between palm and elbow.

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At a later stage, more or less in the early thirties, I also started to see decreased tolerance to heat. I sweat only at the armpits, groin, toes and fingers. My body had really evolved and managed to adapt well to tolerate heat. I can't explain this, but I was amongst the most heat tolerant of the class or football team.

Not sweating heavily, I found advantages like not getting particularly dirty and with an unpleasant smell, or appearance with lots of sweat all over after a bit of exercise. Also, I rarely needed to drink or carry water with me, possibly because I did not lose any water in sweat. However, now that I am getting older, this virtue is lost and is affecting me badly in summer where I cannot tolerate the hot dry months as much as I used to in my young age.

My mother had more or less the same symptoms, perhaps more pronounced as she had lost her hair perhaps faster, and used to complain of heat and feeling lethargic for many summers in her fifties. Later I learnt that she also had unsymmetrical breasts. She also suffered from some paranoia and I don't know if that is related to ED, but I can see that I also have a mild form, perhaps developed over time due to hair loss, and hence you start suspecting at the time your hair is falling visibly and that everyone is looking at you and concerned with your looks. She solved the problem by having a wig.

Now that I am married with a pretty and adorable wife, I do not care that much about the symptoms described above, except the heat intolerance, but this transformation especially at the sensitive teenage to early twenties period, had affected my psychology and I am now very alert, open to worries, easily depressed, and possibly with delusional disorder. If I was a girl, I know that this effect would be much more magnified, since balding for a man is acceptable in today's society but for a girl is a huge shock.

At the moment, my greatest worry is that my daughter, good looking as a model without any exaggeration, would not cope with the shock in this society, always pushing toward perfect physical demands, and in a society that depression is on the increase. My hope is that these symptoms will linger a bit longer until she has established a relationship and have a steady character.

Personally, my worries are if this mutation will bring new conditions later on in my life, as it seems to manifest gradually with new symptoms. But then to think positively, as my wife says, this ED has a number of characters that are in the wish list even of female VIPs: little body hair hence a perfect smooth skin without having to shave; look younger for your age (my 60-year old mum looked as if she was in her late forties!), no sweat-smelly body in hot days, and possibly a long-lived life, unless struck by an unfortunate terminal disease which my mum suffered. Perhaps who knows, my ED contributed to become a fine botanist, where visual perception and visual data processing have to be very acute to identify details and fine morphological differences that are important in natural studies, such high graphical IQ gained to balance the rather below average in literature.

If you would like to know more and have any questions, Stephen has kindly allowed us his email for you to contact him—
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