

ERRATA / CORRIGENDA / ADDENDUM

PhD Dissertation

“Hereditary Angioedema: Clinical Features and Enzyme Polymorphisms in Hereditary Angioedema with F12 Mutation in Southwestern Spanish Families; Management of Pregnancy and Delivery in Patients with Hereditary Angioedema with C1 Inhibitor Deficiency”

Author: Macarena Carmen Piñero Saavedra

Errata

Location	Error	Correction
Page 20, par.1, line 7	LH	LK
Page 29, par.2, line 7	“...pathological.”	“...pathological (<u>Bork et al., 2020a; Ariano et al., 2020; Bork et al., 2021</u>)”
Page 29, par.4, line 1	nonsense	missense
Page 61, par.2, line 3	progesterone	progestin
Page 61, par.2, line 7	progesterone	progestin
Page 67, par.3, line 2	“...specific questionnaire”	“...specific questionnaire (<u>data collection sheet</u>)”
Page 67, par.3, line 3	“...filled out with...”	“...filled out <u>by physicians</u> with...”
Page 67, par.3, line 5	“The research ethics committees of Hospital Universitario Virgen del Rocío and Hospital Universitario La Paz approved the study.”	“The research ethics committees of Hospital Universitario Virgen del Rocío and Hospital Universitario La Paz (<u>the genetic study was performed in both centers</u>) approved the study.”
Page 68, par.2, line 1	“We reviewed 125 pregnancies in 61 patients as well as 14 miscarriages and 4 induced abortions.”	“We reviewed <u>a total of 143 gestations in 61 patients: 125 full-term pregnancies</u> as well as 14 miscarriages and 4 induced abortions.”
Page 69, par.1, line 6	“pregnancies (71.2%).”	“ <u>full-term pregnancies</u> (71.2%)”
Page 69, par.2, line 2	“the pregnancies...”	“the <u>125 full-term pregnancies</u> ...”
Page 69, par.2, line 5	“of pregnancies...”	“of <u>full-term pregnancies</u> ...”
Page 73, par.2, line 2	“the pregnancies in...”	“the <u>full-term pregnancies</u> in...”
Page 73, par.3, line 3	“...36 pregnancies...”	“...36 <u>full-term pregnancies</u> ...”
Page 74, par.1, line 1	“of the pregnancies...”	“of the <u>full-term pregnancies</u> ...”

Corrigenda / Addendum

Abstract, page ix

Add the following paragraph at the end of the abstract:

“In conclusion, our research has contributed to the knowledge of epidemiological, clinical, treatment and genetic aspects of this rare disease.”

Page 46, Genotyped for SNP -2399A in XPNPEP2

Add the following information below the paragraph “The XPNPEP2 c-2399A polymorphism (chromosome X) was genotyped using a real-time PCR TaqMan assay (Life Technologies, Carlsbad, California) following the manufacturer’s indications”:

Reactions were carried out in a 96 well-plate in the Real-time PCR cycler LightCycler® System (Roche Molecular Systems, Basel, Switzerland). The sequences of the primers used were: forward 5'-GCTCTTCCCCCTGCTGTGT-3', reverse 5'-GGTGCTGTTGGGTGCCTCATC-3'. Amplification reaction contained 12.5 µL of 2 TaqMan Universal Master Mix, 1.0 µL each of 10 µM primers, 0.2 µL of 10 µM probe, and 1.0 µL of template DNA and nuclease-free water to a final volume of 25 µL. The thermal profile consisted of an initial denaturation step for 10 min at 95°C followed by 40 cycles of denaturation (95°C for 15s) and annealing (60°C for 1 min). Fluorescence was measured once per cycle at the end of the 60°C segment. Genotype calls were assessed with LightCycler® Software.

Page 57, Table 2.8. Polymorphisms in ACE gene and XPNPEP2 gene, table legend

Move the sentence indicated in the table legend “No significant differences were found between the expression of symptoms and the presence of [1] combination of (allele I +allele A) vs no combination – p=0.5759 ; [2] D/D vs (I/D; I/I) – p=0.999; [3] C/C or C/- vs A/C or A/- – p=0.999. Fisher’s exact test.” to the end of the main text in that section.

Page 68, Figure 3.1. Distribution of patients and full-term pregnancies among the 5 reference hospitals for hereditary angioedema, table legend

Add “Some patients were followed up in more than one center.” at the end of the table legend.