

**Result certificate #030242:**

**Sample**

Sample: 13-04320  
Name: Jessie B Edens  
Breed: Australian Shepherd  
Reg. number: ASDT-TX-1300063  
Date of birth: 4/6/2010  
Sex: female  
Date received: 04.03.2013  
Sample type: buccal swab

**Detection of g.85286582insC and g.85286582delC mutation in gene HSF4 causing hereditary cataract in several dog breeds by fragment analysis**

**Customer**

Ashley Edens  
315 Synterra Estates Loop  
76458 Jacksboro  
United States

**Result: Mutation was not detected (N/N)**

**Explanation**

Presence or absence of mutation g.85286582delC in HSF4 gene causing hereditary cataract (HC) in Australian Shepherds was tested. Presence of deletion is connected with development of binocular cataract in different age of the dog. Generally, the mutation is inherited in autosomal dominant trait with incomplete penetration. It means that carriers do not need to be affected with HC; there is also possibility involving other genetic or environmental factors.

Individuals with one deleted allele (result N/P, negative/positive) have approximately 17-time higher risk of binocular cataract than the individuals without any deleted allele (result N/N). Heterozygous individuals (N/P) are in higher risk of HC disease and they transfer the mutation to their offspring.

This test does not exclude existence of any other unknown mutation of HSF4 gene nor different gene responsible for hereditary cataract.

Method: SOP25, accredited method

Report date: 08.03.2013

Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

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