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Identification of Two Types of Growth Hormone Receptor Mutations in Two Strains of Sex-linked Dwarf Chickens

[Kenichi Tahara](#)¹⁾, [Akira Tsukada](#)¹⁾, [Takanobu Hanai](#)¹⁾, [Kenta Okumura](#)¹⁾, [Kikumi Yamada](#)¹⁾, [Atsushi Murai](#)¹⁾²⁾, [Rikiya Yamamoto](#)³⁾, [Makoto Maeno](#)⁴⁾, [Noboru Saito](#)¹⁾²⁾ and [Kiyoshi Shimada](#)²⁾

1) Division of Applied Genetics and Physiology, Graduate School of Bioagricultural Sciences, Nagoya University

2) Avian Bioscience Research Center, Graduate School of Bioagricultural Sciences, Nagoya University

3) National Livestock Breeding Center Okazaki Station

4) National Livestock Breeding Center Hyogo Station

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Sex-linked dwarf (SLD) of White Leghorn (WL; S23MA line, Okazaki station, Japan) and White Plymouth Rock (WPR; 15 line, Hyogo station, Japan) strains were established from the MA line (WL) and 16 line (WPR) of a normal growing line, respectively. However, the responsible genes in the two lines of SLD chicken have not been identified. In this study, we characterized the phenotypes and identified the responsible genes in the SLD chickens of these two strains. SLD chickens of both strains showed low body weight, short tarsometatarsus length, and high blood growth hormone (GH) level compared with the normal growing lines. From these particular results, it was indicated that the SLD chickens might possess a defect in the growth hormone receptor (GHR). We identified two types of mutations in the GHR gene in each SLD chicken by Northern blot and polymerase chain reaction analyses. The S23MA line had a single base mutation in the splice donor site of the exon 5/intron 5 on the GHR gene, whereas the 15 line lacked a large part of exon 10 of the GHR gene, which contained 27 highly conserved amino acids at the 3' end of the coding region and 3'-UTR. Furthermore, it was revealed that growth retardation was caused by reduction in food intake of the SLD chickens. These two genetically distinguishable lines of

dwarf chickens would serve as an effective tool for analyzing novel GH function and GHR signal transduction in chickens.

Keywords: [genotyping](#), [growth hormone](#), [growth hormone receptor](#), [growth hormone resistance](#), [sex-linked dwarf chicken](#)

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