News Release

Title

A new treatment for autosomal recessive woolly hair/hypotrichosis caused by *LIPH* mutations

Key Points

- **We proved that "minoxidil" is effective for autosomal recessive woolly hair/hypotrichosis caused by *LIPH* mutations**

Summary

Prof. Masashi Akiyama, Dr. Tomoki Taki at Department of Dermatology, Nagoya University Graduate School of Medicine, and Prof. Kazumitsu Sugiura at Department of Dermatology, Fujita Health University School of Medicine, evaluated the efficacy and safety of minoxidil for autosomal recessive woolly hair/hypotrichosis (ARWH) caused by *LIPH* mutations in a clinical trial. ARWH is rare diseases and there have been no effective treatments for ARWH. However, minoxidil was effective for all participants in the present study and a half of the patients showed drastic improvement.

Research Background

ARWH is a rare hereditary hair disease characterized by short and tightly curled scalp hair with hypotrichosis. ARWH is known to be mainly caused by mutations in *LIPH*. *LIPH* encodes a membrane-bound member of the mammalian triglyceride lipase family, lipase H (LIPH). LIPH is involved in the pathway of hair differentiation and growth regulation. In the Japanese population, the carrier rate of ARWH-causative *LIPH* mutations is about 2%. This means that carriers of the *LIPH* mutations are by no means rare. Until now, there have been no effective treatment for ARWH, and patients have to accept their hypotrichosis and curly hair as one individuality, or to use a wig. Therefore, a clinical trial to confirm the efficacy of "minoxidil" against ARWH has been waited eagerly for a long time.

Research Results

This one-year, single-center, open-label, prospective interventional study enrolled patients from the dermatology outpatient clinic of Nagoya University Hospital in Aichi, Japan. 8 cases were enrolled, including 5 children, to evaluate the efficacy and safety of the topical 1% minoxidil lotion (10 mg minoxidil in 1 mL of lotion with propylene glycol as the main ingredient). By the end of the present study, the hypotrichosis of every subject with *LIPH* mutations had improved and 4 of them had shown dramatic improvement (see Figures 1 and 2). There were no serious adverse events, but some mild adverse events (dry skin on the scalp, trichiasis and mild hypertrichosis on the entire body).

Figure1. Clinical features of ARWH patients with *LIPH* mutations before and after topical minoxidil treatment

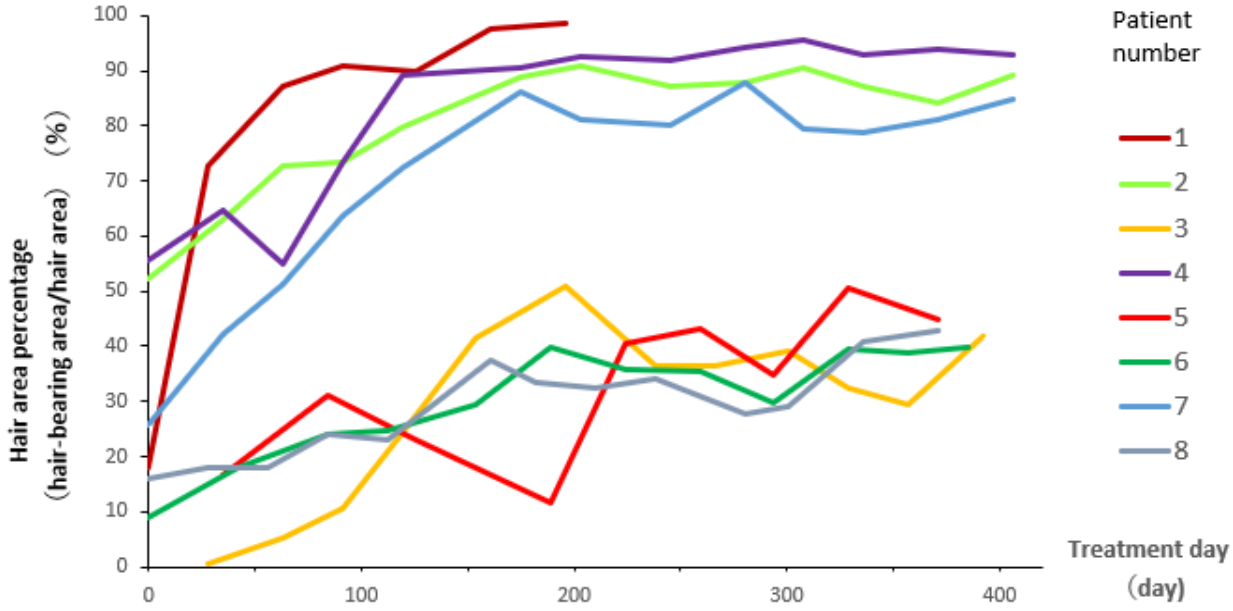

Figure 2. Response of nine hereditary hypotrichosis patients to topical minoxidil treatment

The hypotrichosis of every subject with *LIPH* mutations had improved.

Research Summary and Future Perspective

There have been no effective treatments for ARWH. However, the present prospective interventional study suggests that minoxidil could improve hypotrichosis in ARWH due to *LIPH* mutations. The topical minoxidil therapy is thought to be a promising novel treatment for ARWH.

Publication

Tomoki Taki[1], Kana Tanahashi[1], Takuya Takeichi[1], Takenori Yoshikawa[1], Yuya Murase[1], Kazumitsu Sugiura[2], Masashi Akiyama[1]

1.Department of Dermatology, Nagoya University Graduate School of Medicine, 65 Tsurumai-cho, Showa-ku, Nagoya, Aichi 466-8550, Japan

2.Department of Dermatology, Fujita Health University School of Medicine, 1-98 Dengakugakubo, Kutsukake-cho, Toyoake, Aichi 470-1192, Japan

Association of Topical Minoxidil With Autosomal Recessive Woolly Hair/Hypotrichosis Caused by *LIPH* Pathogenic Variants

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