P0001013 (111222333EP)

Identifier: 111222333EP

Patient name: Doe, John

Life status: Alive

Date of birth: 2019-02-25

Sex: Male

Indication for referral:

Referred by pediatrician for genetics evaluation of congenital sensorineural deafness. Child has bright blue eyes. Mom was noted to have a white forelock. Maternal cousin and uncle with hearing loss.

**Pre-visit questionnaire scheduler**

**Medical history**

Medical and developmental history:

Moderate bilateral sensorineural hearing loss confirmed by auditory brainstem response (ABR) at age 3 months. Hearing-aid fitted at 6 months. Normal language and social development. No known maternal illness in pregnancy and uncomplicated birth.

Allergies - environmental, food, medication:

NKDA

Age of onset:

Congenital onset

**Family history and pedigree**

FAM0000719 (Doe)

Paternal ethnicity:

1. Korean

Maternal ethnicity:

1. Caucasian

2. Irish

List health conditions found in family (describe the relationship with proband)

Maternal cousin and uncle with hearing loss

Other affected relatives

NO Consanguinity

NO Parents with at least 3 miscarriages

**Prenatal and perinatal history**

NO Multiple gestation

Gestation at delivery (weeks) Term birth

PREGNANCY HISTORY

NO Maternal diabetes

NO Maternal fever in pregnancy

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P0001013 (111222333EP)

NO Maternal teratogenic exposure

PRENATAL DEVELOPMENT

NO Intrauterine growth retardation

NO Oligohydramnios

NO Polyhydramnios

DELIVERY

NO Premature birth

NEONATAL GROWTH PARAMETERS

Abnormal birth weight

NO Small for gestational age (<-2SD)

NO Large for gestational age (>+2SD)

Abnormal birth length

NO Small birth length (<-2SD)

NO Large birth length (>+2SD)

PERINATAL COMPLICATIONS

NO Neonatal respiratory distress

**Measurements**

**Date: 03/09/2022**

**Age: 3y 0m**

**Weight:** 18.0 kg 97th pctl (+1.82SD) **Height:** 110.0 cm 100th pctl (+3.66SD)

**BMI:** 14.88 27th pctl (-0.6SD) **Head circumference:** 50.0 cm 43rd pctl (-0.18SD)

**Inner canthal distance:** 3.2 cm 98th pctl (+2.08SD) **Interpupilary distance:** 5.6 cm 99th pctl (+2.18SD)

**Clinical symptoms and physical findings**

CRANIOFACIAL

Broad nasal tip

Telecanthus

EYE DEFECTS

Hypertelorism

Blue irides

EAR DEFECTS

Hearing impairment

Moderate sensorineural hearing impairment

Congenital sensorineural hearing impairment

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P0001013 (111222333EP)

**Genotype information**

LIST OF GENES

**Gene Status Strategy Comments**

1 PAX3 Candidate Sequencing

**Variants in PAX3 (1)**

**# cDNA Interpretation Zygosity**

1.1 c.\*175C>T Likely Pathogenic heterozygous

**Reference genome** GRCh37 (hg19)

**Protein** p.Gln487Ter

**Transcript** NM\_181458.4

**Inheritance** unknown

**Diagnosis**

Final diagnosis (OMIM)

#193500 WAARDENBURG SYNDROME, TYPE 1

Case solved

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b. 1947 (75y)

3

**A&W**

P0001013 (111222333EP)

d. 75y b. 1952 (70y) (heart attack)

b. 1952 (70y)

hearing loss in older age

P

b. Jun 26, 1986 (35y)

Korean

no hearing concerns

**John Doe**

b. Feb 18, 1987 (35y) Caucasian, Irish

tx. with antibiotics in infancy

15 weeks b. 2021 (1y) 10 weeks P0001013

[ 111222333EP ]

b. Feb 25, 2019 (3y)

**Legend:**

b. 2016 (6y)

Phenotypes:

⬤ Blue irides

⬤ Hearing impairment

⬤ Premature graying of hair

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P0001013 (111222333EP)

Weight for age, 2 to 20 years, boys Age (years)

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Age (years)

The WHO Growth Charts for Canada

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Record last modified by Administrator on 2022/03/09 14:58, exported by Administrator.

This is a PDF of the PhenoTips record on 2022/03/09 14:58 which may have since been modified. For the latest record please check: PhenoTips.

P0001013 (111222333EP)

Height for age, 2 to 20 years, boys Age (years)

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Age (years)

The WHO Growth Charts for Canada

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Record last modified by Administrator on 2022/03/09 14:58, exported by Administrator.

This is a PDF of the PhenoTips record on 2022/03/09 14:58 which may have since been modified. For the latest record please check: PhenoTips.

P0001013 (111222333EP)

Body mass index for age, 2 to 20 years, boys Age (years)

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20

Age (years)

The WHO Growth Charts for Canada

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Record last modified by Administrator on 2022/03/09 14:58, exported by Administrator.

This is a PDF of the PhenoTips record on 2022/03/09 14:58 which may have since been modified. For the latest record please check: PhenoTips.

P0001013 (111222333EP)

Head circumference for age, 2 to 20 years, boys Age (years)

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Age (years)

United States Head Circumference Growth Reference Charts: Birth to 21 Years (Holden et al., 2010)

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