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

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

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	

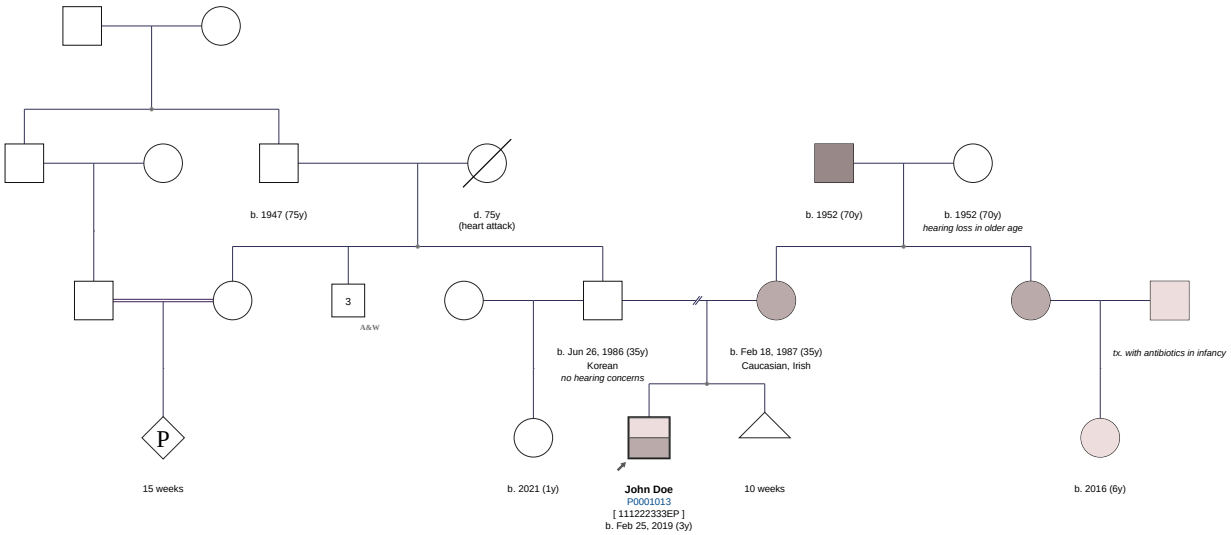
Reference genome	GRCh37 (hg19)
Protein	p.Gln487Ter
Transcript	NM_181458.4
Inheritance	unknown

Diagnosis

Final diagnosis (OMIM)

#193500 WAARDENBURG SYNDROME, TYPE 1

Case solved

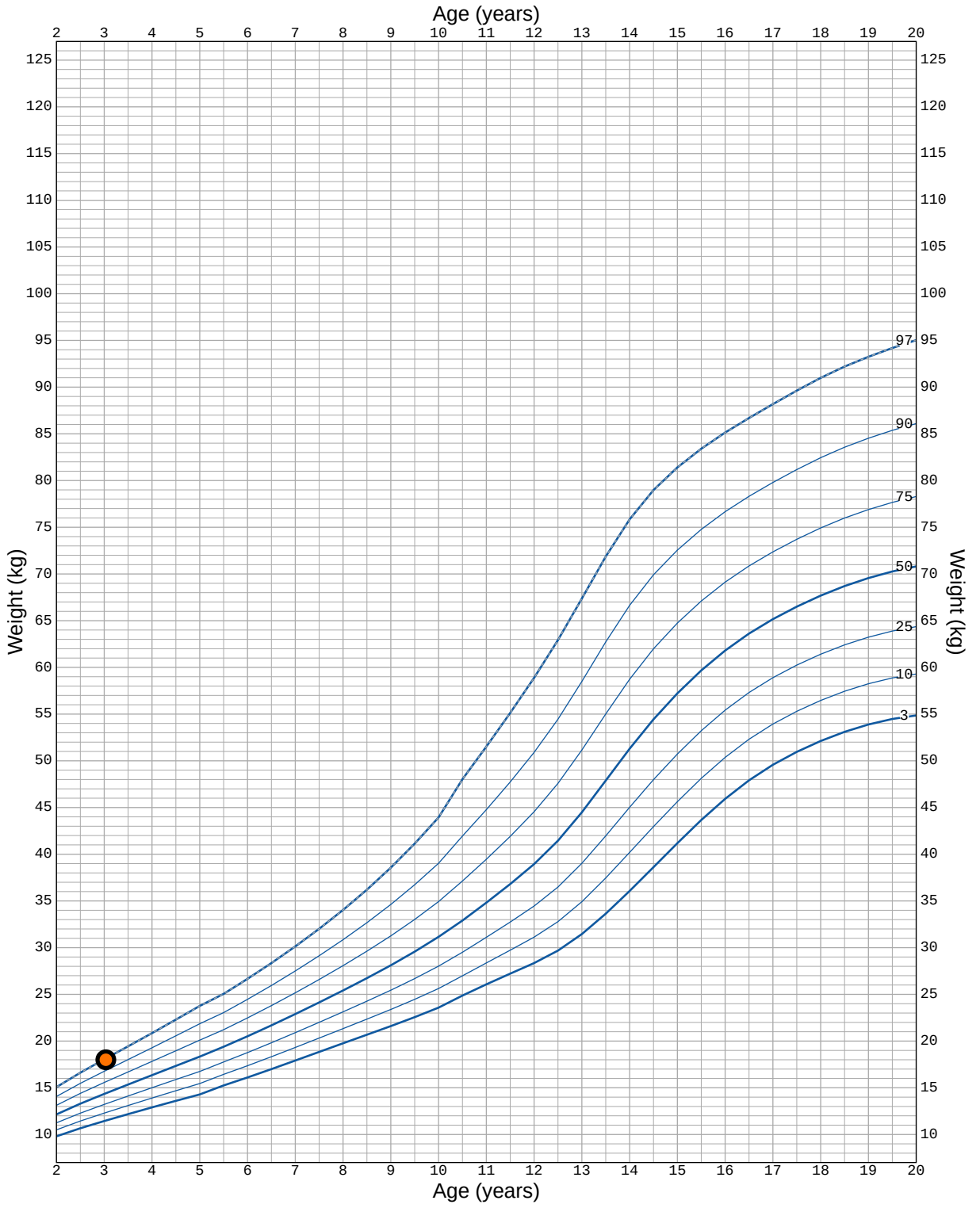


Legend:

Phenotypes:

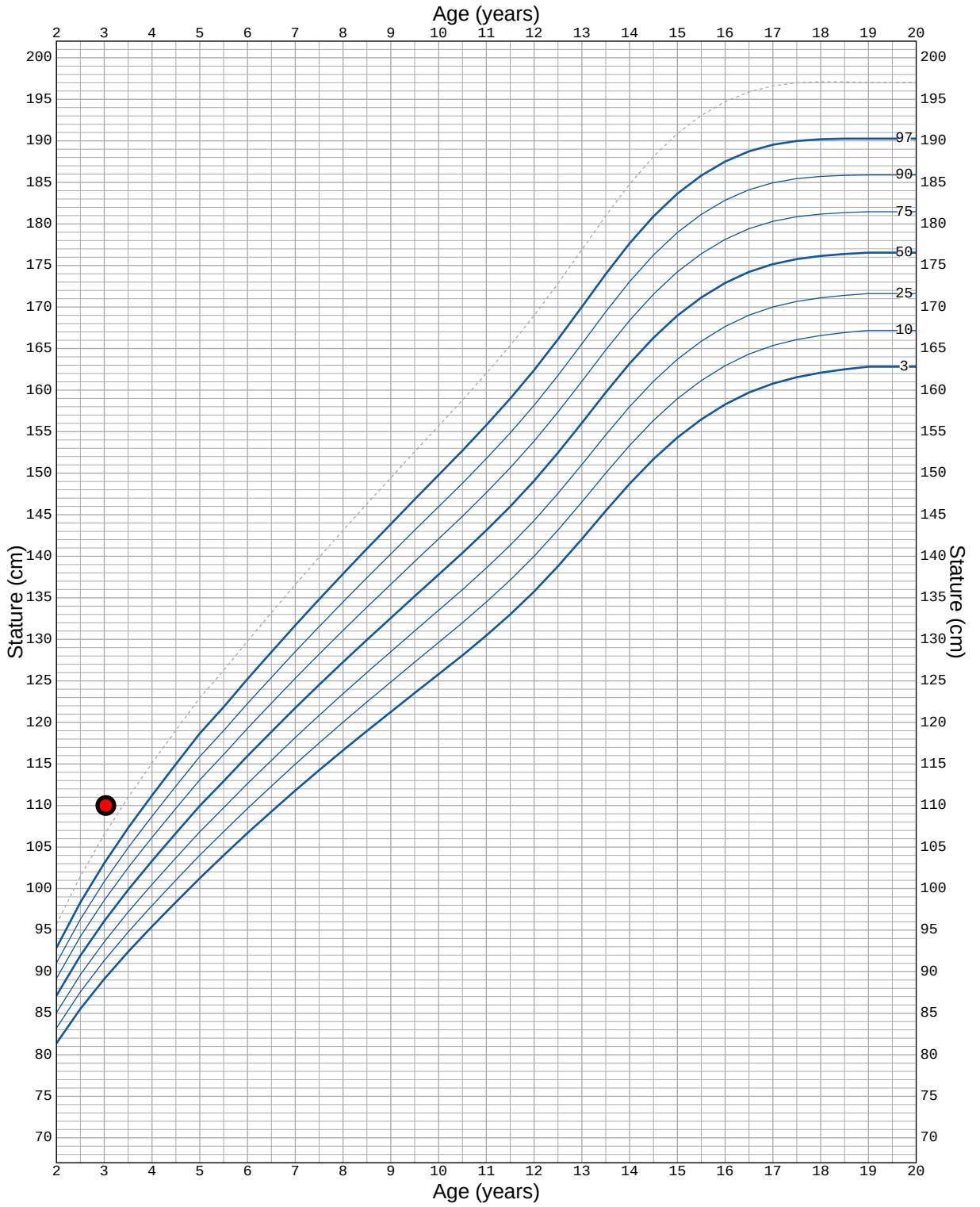
- Blue irides
- Hearing impairment
- Premature graying of hair

Weight for age, 2 to 20 years, boys



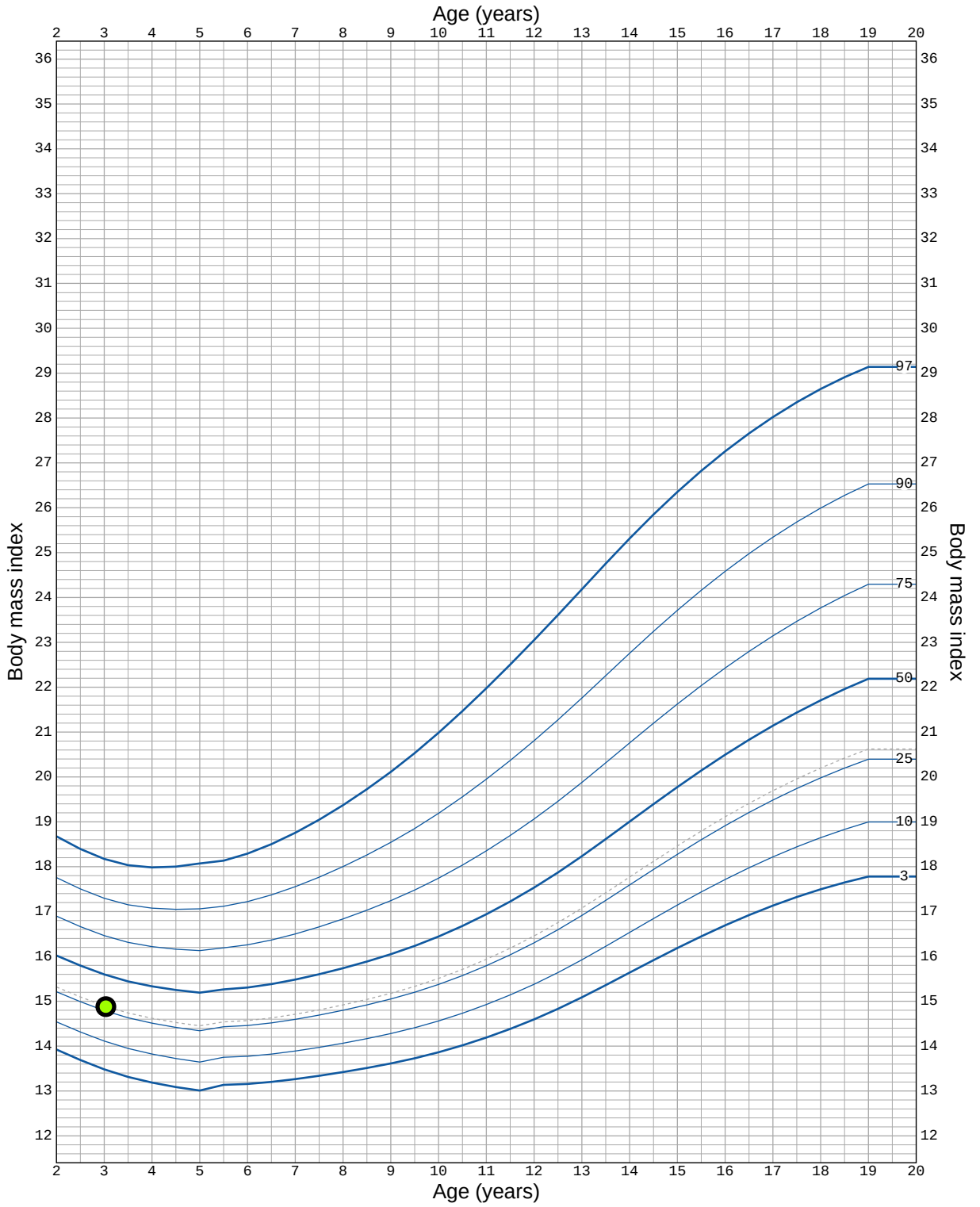
The WHO Growth Charts for Canada

Height for age, 2 to 20 years, boys



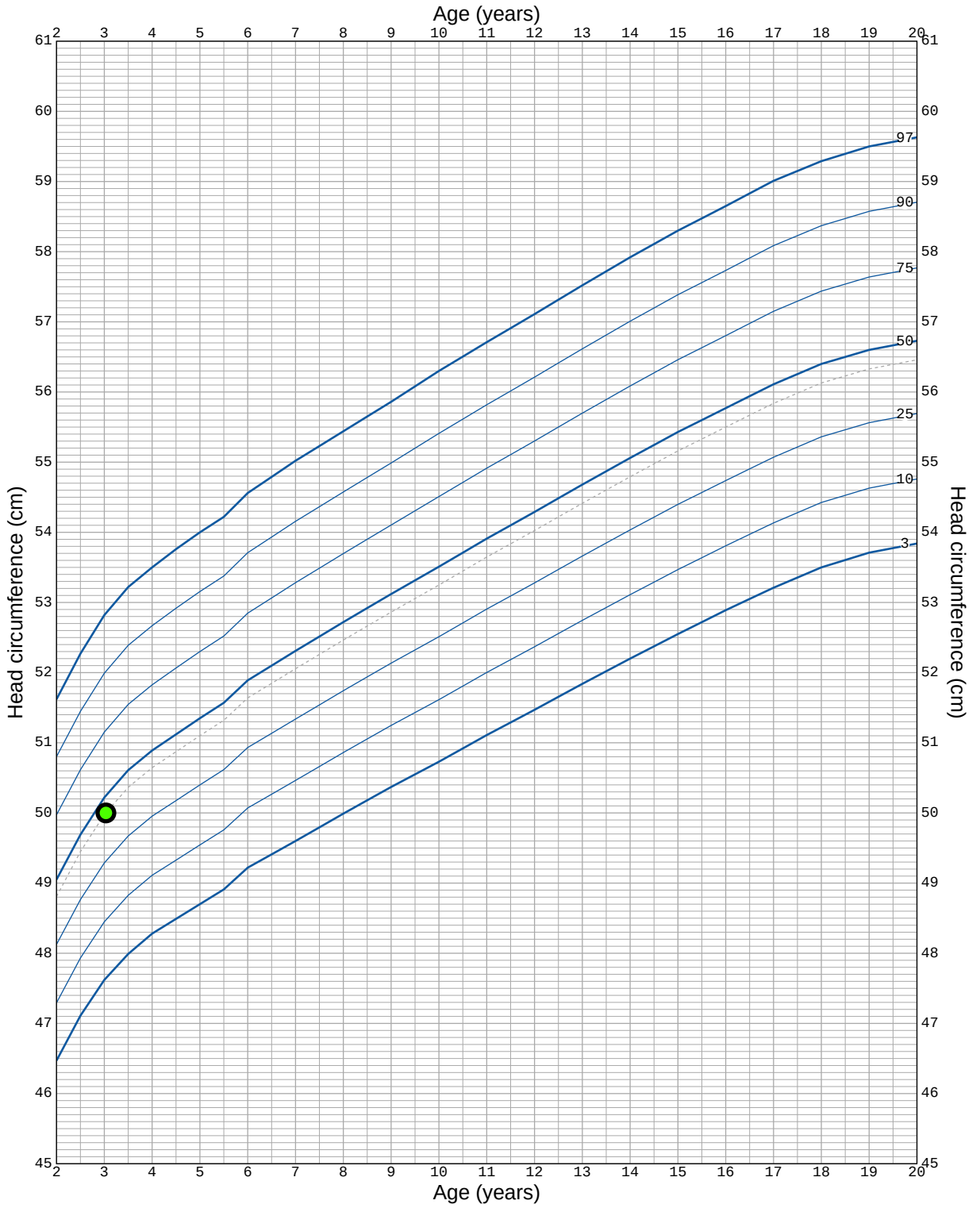
The WHO Growth Charts for Canada

Body mass index for age, 2 to 20 years, boys



The WHO Growth Charts for Canada

Head circumference for age, 2 to 20 years, boys



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