

# General Medical History Toolkit Guideline

## v2.0

### Instruments

#### Toolkit Purpose

A collection of measures to capture essential phenotypes associated with General Medical History (GMH) research.

#### Guideline Description

The GMH toolkit can be used to collect essential phenotypes associated with GMH research, including: medical history related to allergies, pregnancy(ies), surgeries, vision and hearing disorders, mental disorders, respiratory system disorders, nervous system disorders, musculoskeletal system disorders, endocrine system disorders, digestive system disorders, cardiovascular system disorders, urogenital system disorders, circulatory system disorders, integumentary system disorders, sexually transmitted diseases, and developmental disorders. The following document establishes guidelines (particularly applicable in Africa) on how to use the toolkit and collect detailed, relevant and harmonized phenotype and exposure data for research.

As listed below, the GMH toolkit consists of 16 Instruments, labelled Instruments 1 to 16:

<b>Instrument</b>	<b>Phenotypes</b>	<b>Instrument</b>	<b>Phenotypes</b>
1	Vaccinations	9	Respiratory System Disorders
2	Allergies	10	Cardiovascular System Disorders
3	Current Pregnancy	11	Digestive System Disorders
4	Pregnancy History	12	Urogenital System Disorders
5	Last Pregnancy Outcome	13	Endocrine System Disorders
6	Mental Disorders	14	Circulatory System Disorders
7	Nervous System Disorders	15	Musculoskeletal System Disorders
8	Vision & Hearing Disorders	16	Integumentary System Disorders

## Important Notes

1. The toolkit employs branching logic, therefore, we recommend that it is completed in order, as some variables may or may not appear OR accept input based on the input of previously listed variables.
2. Some branching logic (specifically related to date of birth/age and current pregnancy) affects the display of items relevant to adult or paediatric participants across multiple instruments.
3. Any addition or removal of variables may also affect branching logic so editing of variables should be carefully positioned so as not to interrupt branching logic conditions with related variables.
4. The toolkit is recommended to be used in conjunction with the Core Phenotypes toolkit (<https://github.com/h3abionet/h3aphenstds>).
5. Although not highlighted below, each instrument requires a collection date, which can be collected either manually or automatically.
6. Consistent codes are recommended for the identification of missing data, and these are incorporated into all Instruments discussed below.
7. Codes for Missing Data are specified below:

Code	Value Label
-991	No information
-992	Asked but unknown
-993	Temporarily unavailable
-994	Not asked
-995	Refused
-998	Not applicable

8. We recommend that when a participant responds with an "I don't know" to a question that the interviewer firstly ensures that the participant understands the question clearly and secondly is gently encouraged to reconsider their response if possible. If "I don't know" is still the response we make use of the 'Asked but unknown' missing code. Questions where "I don't know" is a highly anticipated and valid response will have a checkbox for Unknown included - it should be noted that this will not be recognised as missing data in statistical software.

## Recommendations

### Instrument 1: Vaccinations

The instrument enables the retrospective self-report collection and(or) recording of vaccination history from research participants or medical records, respectively.

Questions	<p>Has the participant received any of the following vaccinations?</p> <p><b>Response Options:</b></p> <p>Yes; No; Don't Know</p> <p>Vaccinations (See notes)</p> <p>(If Other) Specify other vaccinations received:</p>																	
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	parasites that are transmitted by Anopheles mosquito bites to humans and is typically clinically characterized by attacks of fever, headache, chills and vomiting.
<b>Measles</b>	A highly contagious viral infection caused by the measles virus. Symptoms appear 8-12 days after exposure and include a rash, cough, fever and muscle pains that can last 4-7 days. Measles vaccines are available to provide prophylaxis, usually combined with mumps and rubella vaccines.
<b>Mumps</b>	A contagious viral infection caused by the mumps virus. Symptoms include swollen and tender parotid glands, fever, muscle aches and fatigue.
<b>Pertussis/ Whooping Cough</b>	A contagious bacterial respiratory infection caused by Bordetella pertussis. It is characterized by severe and uncontrollable cough, resulting in a whooping sound during breathing following the cough.
<b>Poliomyelitis</b>	An acute infectious disorder that affects the nervous system. It is caused by the poliovirus. The virus spreads by direct contact, and can be prevented by prophylaxis with the polio vaccine.
<b>Rotavirus</b>	A viral infection, which is the leading cause of severe acute gastroenteritis.
<b>Rubella</b>	A viral infection caused by the rubella virus. It is initially manifested with flu-like symptoms that last one or two days, followed by the development of a characteristic red rash which lasts from one to five days.
<b>Tetanus</b>	A serious infectious disorder that follows wound contamination by the Gram-positive bacterium Clostridium tetani. The bacteria produce a neurotoxin called tetanospasmin, which causes muscle spasm in the jaw and other anatomic sites.
<b>Tuberculosis</b>	A chronic, recurrent infection caused by the bacterium Mycobacterium tuberculosis. Tuberculosis (TB) may affect almost any tissue or organ of the body with the lungs being the most common site of infection.
<b>Typhoid</b>	Typhoid or typhoid fever is a reportable, fecal-oral, potentially fatal infectious disease, caused by the bacteria Salmonella typhi and characterized by a non-focal fever.
<b>Yellow fever</b>	Yellow fever (YF), caused by YF virus, is a zoonotic disease characterized by fever and constitutional symptoms, with the potential to progress to severe and fatal viral hemorrhagic fever with shock and multi-organ system failure.

## Instrument 2: Allergies

The instrument enables the self-report collection and(or) recording of current allergy status from research participants.

Questions	<p>Does the participant have any known allergies?</p> <p><b>Response Options:</b> Yes; No</p> <p>(If Yes) Please specify</p> <p><b>Response Options:</b></p> <p>Milk/Lactose; Eggs; Tree Nuts; Peanuts; Shellfish; Wheat; Soy; Fish; Drug Allergies; Other</p> <p>(If Drug Allergy) Specify which drug/s:</p> <p>(If Other) Specify other allergy type/s:</p>
Notes	<ul style="list-style-type: none"> <li>- <b>Allergy:</b> an immune system response to a foreign substance that's not typically harmful to your body. These foreign substances are called allergens. They can include certain foods, pollen, or pet dander.</li> </ul>

## Instrument 3: Current Pregnancy

The instrument enables the collection of current pregnancy status from research participants.

Questions	<p>Is the participant currently pregnant?</p> <p><b>Response Options:</b> Yes; No</p> <p>(if YES) Estimated due date OR assessed gestational weeks:</p> <p>(if YES) is the number of fetuses known?</p> <p>(if YES) If Yes, number of fetuses:</p>
Notes	<ul style="list-style-type: none"> <li>- <b>Foetus:</b> the unborn baby that develops from an embryo.</li> <li>- <b>Gestational age:</b> a measure of the age of a pregnancy which is taken from the beginning of the woman's last menstrual period.</li> </ul>

## Instrument 4: Pregnancy History

The instrument enables the retrospective self-report collection and(or) recording of pregnancy history from research participants or medical records, respectively.

Questions	<p>Gravida (number of total prior pregnancies):</p> <p>Para gravida (number of total prior pregnancies &gt;20 gest. weeks):</p> <p>Number of total prior vaginal deliveries:</p> <p>Number of total prior caesarean deliveries:</p> <p>Currently breastfeeding?</p> <p><b>Response Options:</b> Yes; No</p>
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Notes	<ul style="list-style-type: none"> <li>- <b>Caesarean delivery:</b> a surgical procedure used to deliver a baby through incisions in the abdomen and uterus.</li> <li>- <b>Vaginal delivery:</b> the delivery of a baby through the vagina.</li> </ul>
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### Instrument 5: Last Pregnancy Outcome

The instrument enables the retrospective self-report collection and(or) recording of the research participant's last pregnancy outcome.

Questions	<p>Date of last pregnancy outcome:  Years since last pregnancy outcome:  Please specify last pregnancy outcome:  <b>Response Options:</b>  Abortion – induced;  Abortion – spontaneous;  Live birth – full term;  Live birth – pre-term (&lt;37 weeks gestation);  Neonatal death (0 – 6 days)  Stillborn</p> <p>Delivery method:  <b>Response Options:</b>  Caesarean;  Unassisted vaginal;  Assisted vaginal delivery - tongs;  Assisted vaginal delivery - suction</p> <p>Number of babies delivered:  Was there a congenital abnormality present?  <b>Response Options:</b> Yes; No  [(If live birth) Weight of baby/ies &lt; 2.5kg?  <b>Response Options:</b> Yes; No  (If live birth) Weight of baby/ies &gt; 4.5kg?  <b>Response Options:</b> Yes; No</p>
Notes	<ul style="list-style-type: none"> <li>- <b>Neonatal death:</b> Death of a live newborn during the first 28 days of life.</li> <li>- <b>Stillborn:</b> the death or loss of a baby before or during delivery.</li> <li>- <b>Congenital abnormality:</b> structural or functional anomalies that occur during intrauterine life and can be identified prenatally, at birth, or sometimes may only be detected later in infancy.</li> <li>- <b>Assisted delivery - tongs:</b> A type of assisted vaginal delivery. It's sometimes needed in the course of vaginal childbirth. In this delivery, a health care provider applies forceps — an instrument shaped like a pair of large spoons or salad tongs — to the baby's head to help guide the baby out of the birth canal.</li> <li>- <b>Assisted delivery - suction:</b> A type of assisted delivery. During a vaginal delivery that has stalled, a doctor will place a suction cup with a handle on the baby's head to help guide her through the birth canal and into the world.</li> </ul>

### Instrument 6: Mental Disorders

The instrument enables the collection and(or) recording of mental disorder status from research participants or medical records, respectively.

Questions	<p>Has the participant ever been diagnosed with a mental disorder?</p> <p><b>Response Options:</b> Yes; No</p> <p>(If Yes) Specify diagnosed mental disorder/s:</p> <p><b>Response Options:</b> See notes (Disease descriptions)</p> <p>(If Other) Specify other diagnosed mental disorder/s:</p>																
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		expressions of anger), pathological gambling and trichotillomania (excessive hair pulling).
	<b>Posttraumatic Stress Disorder (PTSD)</b>	A disorder arising from experiencing or witnessing a terrifying event. The condition may last months or years, with triggers that can bring back memories of the trauma accompanied by intense emotional and physical reactions. Symptoms may include nightmares or flashbacks, avoidance of situations that bring back the trauma, heightened reactivity to stimuli, anxiety, or depressed mood.
	<b>Personality Disorder</b>	A mental disorder in which you have a rigid and unhealthy pattern of thinking, functioning, and behaving. A person with a personality disorder has trouble perceiving and relating to situations and people. This causes significant problems and limitations in relationships, social activities, work and school.
	<b>Substance Abuse Disorder</b>	A disease that affects a person's brain and behaviour and leads to an inability to control the use of a legal or illegal drug or medication
	<b>Tourette Syndrome</b>	Tourette Syndrome (TS) is a condition of the nervous system. TS causes people to have “tics”. Tics are sudden twitches, movements, or sounds that people do repeatedly. People who have tics cannot stop their body from doing these things.

### Instrument 7: Nervous System Disorders

The instrument enables the self-report collection and(or) recording of nervous system disorder status from research participants or medical records, respectively.

Questions	<p>Has the participant ever been diagnosed with any nervous system disorder?</p> <p><b>Response Options:</b> Yes; No</p> <p>(If Yes) Specify diagnosed nervous system disorder/s:</p> <p><b>Response Options:</b> See notes (Disease descriptions)</p> <p>(If Other) Specify other diagnosed nervous system disorder/s:</p>						
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		disability in childhood. Cerebral means having to do with the brain. Palsy means weakness or problems with using the muscles.
	<b>Dementia</b>	A group of thinking and social symptoms that interferes with daily functioning, characterised by impairment of at least two brain functions, such as memory loss and judgement. Symptoms include forgetfulness, limited social skills and thinking abilities so impaired that it interferes with daily functioning.
	<b>Epilepsy</b>	A disorder in which nerve cell activity in the brain is disturbed, causing seizures. Epilepsy may occur as a result of a genetic disorder or an acquired brain injury, such as a trauma or stroke. During a seizure, a person experiences abnormal behaviour, symptoms and sensations, sometimes including loss of consciousness.
	<b>Hydrocephalus</b>	The buildup of fluid in the cavities (ventricles) deep within the brain. The excess fluid increases the size of the ventricles and puts pressure on the brain.
	<b>Motor Neuron Disease</b>	A nervous system disease that weakens muscles and impacts physical function. In this disease, nerve cells break down, which reduces functionality in the muscles that they supply. The cause is unknown. The main symptom is muscle weakness.
	<b>Multiple Sclerosis (MS)</b>	A disease in which the immune system eats away at the protective covering of nerves. In MS, resulting nerve damage disrupts communication between the brain and the body. MS causes many different symptoms, including vision loss, pain, fatigue and impaired coordination. The symptoms, severity and duration can vary from person to person.
	<b>Parkinson's Disease</b>	A disorder of the central nervous system that affects movement, often including tremors. Nerve cell damage in the brain causes dopamine levels to drop, leading to the symptoms of Parkinson's. Parkinson's often starts with a tremor in one hand. Other symptoms are slow movement, stiffness, and loss of balance.
	<b>Peripheral Neuropathy</b>	Conditions that result when nerves that carry messages to and from the brain and spinal cord from and to the rest of the body are damaged or diseased.
	<b>Stroke</b>	Sudden impairment of blood flow to a part of the brain due to occlusion or rupture of an artery to the brain.
	<b>Traumatic Brain Injury</b>	Brain dysfunction caused by an outside force, usually a violent blow to the head.
	<b>Traumatic Spinal Cord Injury</b>	Damage to any part of the spinal cord or nerves at the end of the spinal canal. A traumatic spinal cord injury may happen because of a sudden blow or cut to the spine.

## Instrument 8: Vision &amp; Hearing Disorders

The instrument enables the self-report collection and(or) recording of vision and hearing disorder status from research participants or medical records, respectively.

Questions	<p>Has the participant ever been diagnosed with a disorder which affects their vision or hearing?</p> <p><b>Response Options:</b> Yes; No</p> <p>(If Yes) Please specify</p> <p><b>Response Options:</b> See notes (Disease descriptions)</p> <p>(If Other) Specify other vision or hearing disorder/s:</p>																						
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### Instrument 9: Respiratory System Disorders

The instrument enables the self-report collection and(or) recording of respiratory system disorder status from research participants or medical records, respectively.

Questions	<p>Has the participant ever been diagnosed with any respiratory system disorder?  <b>Response Options:</b> Yes; No;          (If Yes) Specify diagnosed respiratory system Asthma disorder/s:  <b>Response Options:</b> See notes (Disease descriptions)          (If Other) Specify other diagnosed respiratory disorder/s:</p>														
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### Instrument 10: Cardiovascular System Disorders

The instrument enables the self-report collection and(or) recording of cardiovascular system disorder status from research participants or medical records, respectively.

Questions	<p>Has the participant ever been diagnosed with any cardiovascular disorder?</p> <p><b>Response Options:</b> Yes; No</p> <p>(If Yes) Specify diagnosed cardiovascular disorder/s:</p> <p><b>Response Options:</b> See notes (Disease descriptions)</p> <p>(If Other) Specify other diagnosed cardiovascular disorder/s:</p>																				
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		the site of plaque rupture or erosion that partially or completely occludes coronary blood flow.
	<b>Myocardial Infarction / Heart attack (Type 2)</b>	A myocardial infarction (commonly called a heart attack) is an extremely dangerous condition caused by a lack of blood flow to your heart muscle. Type 2 is defined by a rise and fall of cardiac biomarkers and evidence of ischemia without unstable coronary artery disease (CAD), due to a mismatch in myocardial oxygen supply and demand.
	<b>Obesity</b>	A status with BODY WEIGHT that is grossly above the acceptable or desirable weight, usually due to accumulation of excess FATS in the body. The standards may vary with age, sex, genetic or cultural background. In the BODY MASS INDEX, a BMI greater than 30.0 kg/m <sup>2</sup> is considered obese, and a BMI greater than 40.0 kg/m <sup>2</sup> is considered morbidly obese
	<b>Peripheral Vascular Disease</b>	Any disorder affecting blood flow through the veins or arteries outside of the heart.
	<b>Pulmonary Stenosis</b>	A condition characterized by obstruction to blood flow from the right ventricle to the pulmonary artery. This obstruction is caused by narrowing (stenosis) at one or more points from the right ventricle to the pulmonary artery.
	<b>Valve Disease</b>	In heart valve disease, one or more of the valves in the heart doesn't work properly.

### Instrument 11: Digestive System Disorders

The instrument enables the self-report collection and(or) recording of digestive system disorder status from research participants or medical records, respectively.

Questions	Has the participant ever been diagnosed with any digestive system disorder? <b>Response Options:</b> Yes; No (If Yes) Specify diagnosed digestive system disorder/s: <b>Response Options:</b> See notes (Disease descriptions) (If Other) Specify other diagnosed digestive system disorder/s:					
Notes	<div>- Disease descriptions:</div> <table><tr><th>Disease</th><th>Description</th></tr><tr><td><b>Biliary Atresia</b></td><td>A disease of the bile ducts that affects only infants. Bile is a digestive liquid that is made in the liver. It travels through the bile ducts to the small intestine, where it helps digest fats. In biliary atresia, the bile ducts become inflamed and blocked soon after birth.</td></tr></table>		Disease	Description	<b>Biliary Atresia</b>	A disease of the bile ducts that affects only infants. Bile is a digestive liquid that is made in the liver. It travels through the bile ducts to the small intestine, where it helps digest fats. In biliary atresia, the bile ducts become inflamed and blocked soon after birth.
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	<b>Coeliac Disease</b>	A condition where your immune system attacks your own tissues when you eat gluten.
	<b>Diverticulitis</b>	The infection or inflammation of pouches that can form in your intestines. These pouches are called diverticula.
	<b>Gallstones</b>	The impairment of bile flow, gallstones in the biliary tract, infections, and neoplasms
	<b>Gastroesophageal Reflux Disease</b>	A digestive disorder that occurs when acidic stomach juices, or food and fluids back up from the stomach into the esophagus. GERD affects people of all ages—from infants to older adults.
	<b>Hemorrhoids</b>	Swollen and inflamed veins in the rectum and anus that cause discomfort and bleeding. Haemorrhoids are usually caused by straining during bowel movements, obesity or pregnancy. Discomfort is a common symptom, especially during bowel movements or when sitting. Other symptoms include itching and bleeding.
	<b>Hepato-Splenomagaly</b>	A condition that causes swelling and enlargement of the liver and spleen.
	<b>Hiatus Hernia</b>	A condition in which part of the stomach pushes up through the diaphragm muscle.
	<b>Hirschsprung's Disease</b>	A condition that affects the large intestine (colon) and causes problems with passing stool. The condition is present at birth (congenital) as a result of missing nerve cells in the muscles of the baby's colon.
	<b>Inflammatory Bowel Disease</b>	A group of conditions that cause the digestive system to become inflamed (red, swollen, and sometimes painful). The most common types of IBD are ulcerative colitis and Crohn's disease. These cause similar symptoms, including diarrhea, abdominal pain, and fever.
	<b>Liver Disease</b>	Pathological processes of the liver. Symptoms may include, yellowish skin and eyes (jaundice), abdominal pain swelling, swelling in the legs and ankle, dark urine color, pale stool color and chronic fatigue.
	<b>Malabsorption Syndrome</b>	A number of disorders in which the small intestine can't absorb enough of certain nutrients and fluids.
	<b>Necrotizing Enterocolitis</b>	A disease that affects mostly the intestine of premature infants. The wall of the intestine is invaded by bacteria, which cause local infection and inflammation that can ultimately destroy the wall of the bowel
	<b>Pancreatic Disease</b>	An inflammation of the pancreas. Pancreatitis may start suddenly and last for days or it can occur over many years. It has many causes, including gallstones and chronic, heavy alcohol

		use. Symptoms include upper abdominal pain, nausea and vomiting.
	<b>Peptic Ulcers</b>	Open sores that develop on the inside lining of your stomach and the upper portion of your small intestine. The most common symptom of a peptic ulcer is stomach pain.

## Instrument 12: Urogenital System Disorders

The instrument enables the self-report collection and(or) recording of urogenital system disorder status from research participants or medical records, respectively.

Questions	Has the participant ever been diagnosed with any urogenital system disorder? <b>Response Options:</b> Yes; No (If Yes) Specify diagnosed urogenital system disorder/s: <b>Response Options:</b> See notes (Disease descriptions) (If Yes) Specify other diagnosed urogenital system disorder/s:															
Notes	<div>- Disease descriptions:</div> <table><tr><td>Disease</td><td>Description</td></tr><tr><td><b>Chronic Kidney Disease</b></td><td>A condition characterized by a gradual loss of kidney function over time.</td></tr><tr><td><b>Inguinal Hernia</b></td><td>Also known as groin hernia, occurs when tissue, such as part of the intestine, protrudes through a weak spot in the abdominal muscles. The resulting bulge can be painful, especially when you cough, bend over or lift a heavy object.</td></tr><tr><td><b>Kidney Stones</b></td><td>Hard deposits made of minerals and salts that form inside your kidneys. Diet, excess body weight, some medical conditions, and certain supplements and medications are among the many causes of kidney stones.</td></tr><tr><td><b>Urinary Incontinence</b></td><td>The involuntary leakage of urine. It means a person urinates when they do not want to. Control over the urinary sphincter is either lost or weakened.</td></tr><tr><td><b>Urethral stricture</b></td><td>Involves scarring that narrows the tube that carries urine out of your body (urethra). A stricture restricts the flow of urine from the bladder and can cause a variety of medical problems in the urinary tract, including inflammation or infection.</td></tr><tr><td><b>Urinary Retention</b></td><td>A condition in which you cannot empty all the urine from your bladder. Urinary retention can be acute—a sudden inability to urinate, or chronic—a gradual inability to completely empty the bladder of urine.</td></tr></table>		Disease	Description	<b>Chronic Kidney Disease</b>	A condition characterized by a gradual loss of kidney function over time.	<b>Inguinal Hernia</b>	Also known as groin hernia, occurs when tissue, such as part of the intestine, protrudes through a weak spot in the abdominal muscles. The resulting bulge can be painful, especially when you cough, bend over or lift a heavy object.	<b>Kidney Stones</b>	Hard deposits made of minerals and salts that form inside your kidneys. Diet, excess body weight, some medical conditions, and certain supplements and medications are among the many causes of kidney stones.	<b>Urinary Incontinence</b>	The involuntary leakage of urine. It means a person urinates when they do not want to. Control over the urinary sphincter is either lost or weakened.	<b>Urethral stricture</b>	Involves scarring that narrows the tube that carries urine out of your body (urethra). A stricture restricts the flow of urine from the bladder and can cause a variety of medical problems in the urinary tract, including inflammation or infection.	<b>Urinary Retention</b>	A condition in which you cannot empty all the urine from your bladder. Urinary retention can be acute—a sudden inability to urinate, or chronic—a gradual inability to completely empty the bladder of urine.
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<b>Urinary Retention</b>	A condition in which you cannot empty all the urine from your bladder. Urinary retention can be acute—a sudden inability to urinate, or chronic—a gradual inability to completely empty the bladder of urine.															

## Instrument 13: Endocrine System Disorders

The instrument enables the self-report collection and(or) recording of endocrine system disorder status from research participants or medical records, respectively.

Questions	<p>Has the participant ever been diagnosed with any endocrine system disorder?  <b>Response Options:</b> Yes; No          (If Yes) Specify diagnosed endocrine system disorder/s:  <b>Response Options:</b> See notes (Disease descriptions)          (If Yes) Specify diagnosed endocrine system disorder/s:</p>	
	<ul style="list-style-type: none"> <li>- Listed diseases differ between adult and pediatric participants.</li> <li>- Disease descriptions:</li> </ul>	
	Disease	Description
	<b>Cushing's disorder</b>	A condition that occurs from exposure to high cortisol levels for a long time. The most common cause is the use of steroid drugs, but it can also occur from overproduction of cortisol by the adrenal glands.
	<b>Growth Hormone Deficiency</b>	Also known as dwarfism or pituitary dwarfism, a condition caused by insufficient amounts of growth hormone in the body.
	<b>Hyperthyroidism</b>	Overactivity of the thyroid gland resulting in overproduction of thyroid hormone and increased metabolic rate. The symptoms are related to the increased metabolic rate and include weight loss, fatigue, heat intolerance, excessive sweating, diarrhea, tachycardia, insomnia, muscle weakness, and tremor.
	<b>Hypogonadism</b>	A failure of the gonads, testes in men and ovaries in women, to function properly.
	<b>Hypothyroidism</b>	A condition in which the thyroid gland doesn't produce enough thyroid hormone. Hypothyroidism is a deficiency of thyroid hormones that can disrupt such things as heart rate, body temperature and all aspects of metabolism. Major symptoms include fatigue, cold sensitivity, constipation, dry skin and unexplained weight gain.
	<b>Pituitary adenoma</b>	Non-cancerous tumours in the pituitary gland that don't spread beyond the skull.
	<b>Polycystic ovary syndrome</b>	A hormonal disorder common among women of reproductive age.
<b>Type I Diabetes</b>	A chronic condition in which the pancreas produces little or no insulin. Type I diabetes mellitus is manifested by the sudden onset of severe hyperglycemia with rapid progression to diabetic ketoacidosis unless treated with insulin.	
<b>Type II Diabetes</b>	A type of diabetes mellitus initially characterized by insulin resistance and hyperinsulinemia and subsequently by glucose intolerance and hyperglycemia.	



## Instrument 14: Circulatory System Disorders

The instrument enables the self-report collection and(or) recording of circulatory system disorder status from research participants or medical records, respectively.

Questions	<p>Has the participant ever been diagnosed with any circulatory system disorder?</p> <p><b>Response Options:</b></p> <p>Yes; No</p> <p>(If Yes) Specify diagnosed circulatory system disorder/s?</p> <p><b>Response Options:</b> See notes (Disease descriptions)</p> <p>(If Other) Specify other diagnosed circulatory system disorder/s?</p>																
Notes	<ul style="list-style-type: none"> <li>- Listed diseases differ between adult and pediatric participants.</li> <li>- Disease descriptions:</li> </ul> <table> <tr> <th>Disease</th><th>Description</th></tr> <tr> <td><b>Anemia</b></td><td>A condition in which you lack enough healthy red blood cells to carry adequate oxygen to your body's tissues. Having anemia can make you feel tired and weak. There are many forms of anemia, each with its own cause. Anemia can be temporary or long term, and it can range from mild to severe.</td></tr> <tr> <td><b>Bleeding Disorder</b></td><td>Bleeding disorders are a group of conditions that result when the blood cannot clot properly, resulting in excessive bleeding.</td></tr> <tr> <td><b>Haemophilia</b></td><td>A mostly inherited genetic disorder that impairs the body's ability to make blood clots, a process needed to stop bleeding. This results in people bleeding for a longer time after an injury, easy bruising, and an increased risk of bleeding inside joints or the brain.</td></tr> <tr> <td><b>Podoconiosis</b></td><td>A chronic inflammatory, geochemical skin disease caused by prolonged exposure to irritant red clay soils derived from volcanic rocks and causes bilateral asymmetrical swelling of the lower legs.</td></tr> <tr> <td><b>Rheumatoid Arthritis</b></td><td>An autoimmune disease that can cause joint pain and damage throughout your body. The joint damage that RA causes usually happens on both sides of the body. So, if a joint is affected in one of your arms or legs, the same joint in the other arm or leg will probably be affected, too.</td></tr> <tr> <td><b>SCID</b></td><td>An inherited primary immunodeficiency disease that typically presents in infancy results in profound immune deficiency condition resulting in a weak immune system that is unable to fight off even mild infections.</td></tr> <tr> <td><b>Sickle Cell Disease</b></td><td>A pleiotropic inherited disorder of the blood, characterised by the appearance of sickle-shaped red blood cells and anemia.</td></tr> </table>	Disease	Description	<b>Anemia</b>	A condition in which you lack enough healthy red blood cells to carry adequate oxygen to your body's tissues. Having anemia can make you feel tired and weak. There are many forms of anemia, each with its own cause. Anemia can be temporary or long term, and it can range from mild to severe.	<b>Bleeding Disorder</b>	Bleeding disorders are a group of conditions that result when the blood cannot clot properly, resulting in excessive bleeding.	<b>Haemophilia</b>	A mostly inherited genetic disorder that impairs the body's ability to make blood clots, a process needed to stop bleeding. This results in people bleeding for a longer time after an injury, easy bruising, and an increased risk of bleeding inside joints or the brain.	<b>Podoconiosis</b>	A chronic inflammatory, geochemical skin disease caused by prolonged exposure to irritant red clay soils derived from volcanic rocks and causes bilateral asymmetrical swelling of the lower legs.	<b>Rheumatoid Arthritis</b>	An autoimmune disease that can cause joint pain and damage throughout your body. The joint damage that RA causes usually happens on both sides of the body. So, if a joint is affected in one of your arms or legs, the same joint in the other arm or leg will probably be affected, too.	<b>SCID</b>	An inherited primary immunodeficiency disease that typically presents in infancy results in profound immune deficiency condition resulting in a weak immune system that is unable to fight off even mild infections.	<b>Sickle Cell Disease</b>	A pleiotropic inherited disorder of the blood, characterised by the appearance of sickle-shaped red blood cells and anemia.
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	<b>Systemic Lupus Erythematosus</b>	An autoimmune disease. In this disease, the immune system of the body mistakenly attacks healthy tissue. It can affect the skin, joints, kidneys, brain, and other organs.
	<b>Thalassemia</b>	A blood disorder passed down through families (inherited) in which the body makes an abnormal form or inadequate amount of hemoglobin.
	<b>Thrombocytopenia</b>	A condition in which you have a low blood platelet count. Platelets (thrombocytes) are colorless blood cells that help blood clot. Platelets stop bleeding by clumping and forming plugs in blood vessel injuries.
	<b>Thrombophilia</b>	A condition in which there's an imbalance in naturally occurring blood-clotting proteins, or clotting factors. This can put you at risk of developing blood clots.

### Instrument 15: Musculoskeletal System Disorders

The instrument enables the self-report collection and(or) recording of musculoskeletal system disorder status from research participants or medical records, respectively.

Questions	<p>Has the participant ever been diagnosed with any musculoskeletal system disorder?</p> <p><b>Response Options:</b> Yes; No</p> <p>(If Yes) Specify diagnosed musculoskeletal system disorder/s:</p> <p><b>Response Options:</b> See notes (Disease descriptions)</p> <p>(If Other) Specify other diagnosed musculoskeletal system disorder/s:</p>											
Notes	<div><div><ul style="list-style-type: none"><li>- Listed diseases differ between adult and pediatric participants.</li><li>- Disease descriptions:</li></ul></div><table><tr><th>Disease</th><th>Description</th></tr><tr><td><b>Arthritis</b></td><td>The swelling and tenderness of one or more of your joints. The main symptoms of arthritis are joint pain and stiffness, which typically worsen with age.</td></tr><tr><td><b>Bone Fracture</b></td><td>A medical condition in which there is a partial or complete break in the continuity of the bone.</td></tr><tr><td><b>Carpal Tunnel</b></td><td>Carpal Tunnel is a narrow passageway surrounded by bones and ligaments on the palm side of your hand. When the median nerve is compressed, the symptoms can include numbness, tingling and weakness in the hand and arm.</td></tr><tr><td><b>Fibromyalgia</b></td><td>A disorder characterized by widespread musculoskeletal pain accompanied by fatigue, sleep, memory and mood issues. Researchers believe that fibromyalgia amplifies painful sensations by affecting the way your brain and spinal cord process painful</td></tr></table></div>		Disease	Description	<b>Arthritis</b>	The swelling and tenderness of one or more of your joints. The main symptoms of arthritis are joint pain and stiffness, which typically worsen with age.	<b>Bone Fracture</b>	A medical condition in which there is a partial or complete break in the continuity of the bone.	<b>Carpal Tunnel</b>	Carpal Tunnel is a narrow passageway surrounded by bones and ligaments on the palm side of your hand. When the median nerve is compressed, the symptoms can include numbness, tingling and weakness in the hand and arm.	<b>Fibromyalgia</b>	A disorder characterized by widespread musculoskeletal pain accompanied by fatigue, sleep, memory and mood issues. Researchers believe that fibromyalgia amplifies painful sensations by affecting the way your brain and spinal cord process painful
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		and non-painful signals.
	<b>Gout</b>	A condition caused by a build-up of uric acid usually affecting your feet. Symptoms include swelling and pain in the joints of your foot, particularly your big toe. Sudden and intense pain.
	<b>Guillain Barre syndrome</b>	A rare disorder in which your body's immune system attacks your nerves. Weakness and tingling in your extremities are usually the first symptoms. These sensations can quickly spread, eventually paralyzing your whole body.
	<b>Ligament Injury</b>	A ligament injury usually occurs due to extreme force to a joint such as with a fall or another high-impact event. Common ligament injuries are to the ankle, knee, wrist, thumb, neck, or back ligaments.
	<b>Muscular Dystrophy</b>	A group of inherited diseases that damage and weaken your muscles over time. This damage and weakness is due to the lack of a protein called dystrophin, which is necessary for normal muscle function. The absence of this protein can cause problems with walking, swallowing, and muscle coordination.
	<b>Osteoporosis</b>	A bone disease that occurs when the body loses too much bone, makes too little bone, or both. As a result, bones become weak and may break from a fall or, in serious cases, from sneezing or minor bumps.
	<b>Rickets</b>	A softening and weakening of bones in children, usually due to inadequate vitamin D.
	<b>Scoliosis</b>	Scoliosis is a sideways curvature of the spine that occurs most often during the growth spurt just before puberty.
	<b>Tendonitis</b>	inflammation of a tendon. It happens when a person overuses or injures a tendon, for example, during sport. It is normally linked to an acute injury with inflammation. It often affects the elbow, wrist, finger, thigh, and other parts of the body.

### Instrument 16: Integumentary System Disorders

The instrument enables the self-report collection and/or recording of integumentary system disorder status from research participants or medical records, respectively.

Questions	<p>Has the participant ever been diagnosed with any integumentary disorder?</p> <p><b>Response Options:</b> Yes; No</p> <p>(If Yes) Specify diagnosed integumentary disorder/s:</p> <p><b>Response Options:</b> See notes (Disease descriptions)</p> <p>(If Other) Specify other diagnosed integumentary</p>
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Disease	Description
<b>Albinism</b>	A rare group of genetic disorders that cause the skin, hair, or eyes to have little or no color. Albinism is also associated with vision problems.
<b>Atopic Dermatitis</b>	A chronic inflammatory genetically determined disease of the skin marked by increased ability to form reagin, with increased susceptibility to allergic rhinitis and asthma, and hereditary disposition to a lowered threshold for pruritus. It is manifested by lichenification, excoriation, and crusting, mainly on the flexural surfaces of the elbow and knee.
<b>Eczema</b>	Eczema, also called atopic dermatitis, is a common skin condition marked by itchy and inflamed patches of skin.
<b>Haemangioma</b>	A birthmark often appearing as a rubbery, bright red nodule of extra blood vessels. Haemangiomas are seen more frequently among premature infants.
<b>Neurofibromatosis</b>	A condition characterized by changes in skin coloring (pigmentation) and the growth of tumors along nerves in the skin, brain, and other parts of the body
<b>Psoriasis</b>	Psoriasis is a skin disease that causes red, itchy scaly patches, most commonly on the knees, elbows, trunk and scalp. Psoriasis is a common, long-term (chronic) disease with no cure. It tends to go through cycles, flaring for a few weeks or months, then subsiding for a while or going into remission.
<b>Vitiligo</b>	A condition in which the skin loses its pigment cells. This can result in discolored patches in different areas of the body, including the skin, hair and mucous membranes. Vitiligo is a disease that causes loss of skin color in patches.

## Abbreviations

ADHD: Attention Deficit Hyperactivity Disorder

AIDS: Acquired Immunodeficiency Syndrome

BMI: Body Mass Index

CHF: Congestive Heart Failure

COPD: Chronic Obstructive Pulmonary Disease

COVID-19: Coronavirus 2019

EKG: Electrocardiogram

FH: Family History

GMH: General Medical History

HIV: Human Immunodeficiency Virus

HPV: Human papillomavirus

MS: Multiple Sclerosis

PCO: Polycystic Ovary

PTSD: Post-traumatic Stress Disorder

SCD: Sickle Cell Disease

SCID: Severe Combined Immunodeficiency

STD: Sexually Transmitted Disease

TB: Tuberculosis

TBI: Traumatic Brain Injury

## Administration

### Mode of Administration

	Instruments															
	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16
Interview OR Self-administered questionnaire	X	X	X	X	X	X	X	X	X	X	X	X	X	X	X	X
Clinical assessment																
Bioassay/Lab- based assessment																

### Life Stage

	Instruments															
	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16
Infancy (0 - 12 months)	X	X					X	X	X	X	X	X	X	X	X	X
Toddler (13 - 24 months)	X	X					X	X	X	X	X	X	X	X	X	X
Childhood (2-11 years)	X	X				X	X	X	X	X	X	X	X	X	X	X
Adolescence (12 - 18 years)	X	X	X	X	X	X	X	X	X	X	X	X	X	X	X	X
Adult (18 and older)	X	X	X	X	X	X	X	X	X	X	X	X	X	X	X	X

### Personnel and Training Required

**Instruments 1 to 16** may be implemented as either self-reported questionnaires or interviewer-administered questionnaires. If interviewer-administered, interviews should be conducted by trained or study coordinators or data collectors who speak the native/local language of the target

population. Information in **All Instruments** may also be recorded from hospital and(or) patient records.

## Contributors

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## Contact Us

For queries related to this standard and guideline, users can log a ticket to the Phenotypes Standards queue in the [H3ABioNet Helpdesk](#). User feedback and improvements on the current toolkit are welcome and encouraged. These can also be submitted through the Helpdesk, or on our [GitHub Issues page](#).