The Office of the National Coordinator for Health Information Technology

Synthea™ Module Companion Guide

SPINA BIFIDA

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Prepared by Clinovations Government + Health for the Office of the National Coordinator for Health Information Technology and approved for public release

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Introduction

Synthea™ is an open-source, synthetic patient generator, created by MITRE, that models the medical history of synthetic patients. Clinical disease modules are created using a combination of clinical care protocols and publicly available disease incidence and prevalence statistics. Synthea uses these modules to generate individual synthetic patient records, simulating the progression and treatment of disease from birth to death. Synthea Module Companion Guides serve to orient users to a specific Synthea module. The intended audience includes those who are reviewing a module under development and/or are interested in utilizing the module to generate synthetic patient data.

This document summarizes the scope and intent of the Spina Bifida module. It provides details of the module states and contains a full list of references and data sources used to develop the module.

Module Description

Table 1: Spina Bifida Module Metadata contains a list of metadata attributes that help describe the module, including but not limited to module steward, module developer, date of last update, and other descriptive information.

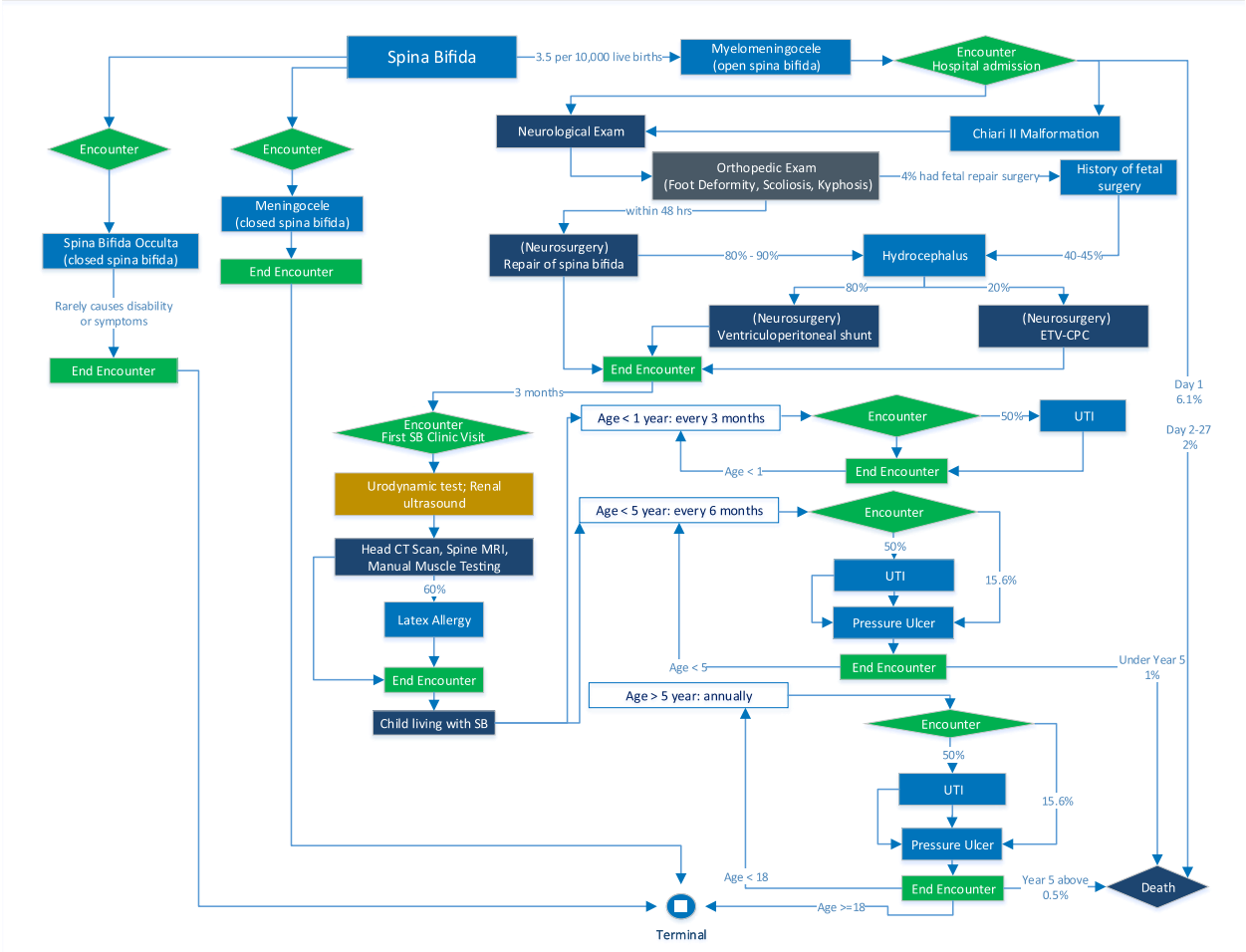
Table 1: Spina Bifida Module Metadata

Metadata	Description
Title	Spina Bifida
Module File Name	spina_bifida
Version Number	1.0
Last Updated	November 30, 2020
Module Steward	Office of the National Coordinator for Health Information Technology (ONC)
Module Developer	Clinovations Government + Health
Description	Spina bifida is the most common, permanently disabling birth defect associated with life.(1) It is a neural tube defect that affects the spine. The three most common types of spina bifida are myelomeningocele, meningocele, and spina bifida occulta. This module focuses on modeling myelomeningocele, the most severe form of spina bifida, for patients age < 18. Myelomeningocele is also known as “open spina bifida.”
Disclaimer	Synthea™ is an open-source synthetic patient generator, created by MITRE, that models the medical history of synthetic patients. This module is developed using the Synthea Module Builder and is limited to the capabilities of Synthea and the Synthea Module Builder. This Synthea module is not a clinical guideline, does not establish a standard of medical care, and has not been tested for all potential applications. THIS MODULE IS PROVIDED "AS IS" WITHOUT WARRANTY OF ANY KIND.
Related Module(s)	none
Reference(s)	Spina Bifida Association Care Coordination Guideline (2) Centers for Disease Control and Prevention (CDC) Spina Bifida (3)

Module Diagram

A Synthea™ module diagram within the Synthea Module Builder is often large and complex to view, as it includes both clinical states and control states. It may be challenging for users to understand and navigate the module within Synthea, especially those who are new to the process. The purpose of the following Visio diagram is to provide a high-level, simplified view of the module contents and flow so users understand the scope and main components of the module before diving into details.

Figure 1: Spina Bifida Visio Diagram



Module States

Table 2: Spina Bifida Module States provides details about each clinical and control state modeled within the module. State Names are modeled in the Spina Bifida module. The Type column indicates the Synthea state type used to define the state. State Remarks provide detailed documentation for each state, including notes, references, and data sources used to define probabilities. The Terminology column identifies the standard codes used to model the clinical states.

Table 2: Spina Bifida Module States

State Name	Type	State Remarks	Terminology
Initial	Initial	Initial state of a module required by Synthea™.	n/a
Enter_Spina_Bifida	Simple	This is a simple state that transitions to the three common types of spina bifida.	n/a
Myelomeningocele	ConditionOnset	0.035% of newborns are born with myelomeningocele. (3) (4) (5) Diagnosed at Encounter_NICU	System: SNOMED-CT Code: 414667000 Display: Meningomyelocele (disorder)
Meningocele	ConditionOnset	0.01% of newborns are born with meningocele. Diagnosed at Encounter_Meningocele	System: SNOMED-CT Code: 171131006 Display: Meningocele (disorder)
Spina_Bifida_Occulta	ConditionOnset	0.025% of newborns are born with Spina Bifida Occulta. Diagnosed at Encounter_Occulta	System: SNOMED-CT Code: 76916001 Display: Spina bifida occulta (disorder)
Encounter_NICU	Encounter	Newborn with myelomeningocele admitted to NICU. Reason: Myelomeningocele	System: SNOMED-CT Code: 32485007 Display: Hospital admission (procedure)
Chiari_II_Malformation	ConditionOnset	90% of myelomeningocele patients also have Chiari_II_Malformation. (6) Diagnosed at Encounter_NICU	System: SNOMED-CT Code: 203082005 Display: Fibromyalgia (disorder)

State Name	Type	State Remarks	Terminology
Neurological_Exam	Procedure	All newborns with myelomeningocele receive a neurological exam. Reason: Myelomeningocele	System: SNOMED-CT Code: 84728005 Display: Neurological examination (procedure)
Orthopedic_Exam	Procedure	All newborns with myelomeningocele receive an orthopedics exam. Reason: Myelomeningocele	System: SNOMED-CT Code: 36321500 Display: Musculoskeletal system physical examination (procedure)
Foot_Deformity	Condition Onset	80-95% of newborns with myelomeningocele have foot deformities.(7) Set the probability to 90%. Diagnosed at Encounter_NICU	System: SNOMED-CT Code: 302297009 Display Congenital deformity of foot (disorder)
No_Foot_Deformity	Simple	This is a simple state for transition to the path of myelomeningocele newborns without foot deformities.	n/a
Scoliosis	Condition Onset	53% of myelomeningocele newborns have scoliosis. (8) Diagnosed at Encounter_NICU	System: SNOMED-CT Code: 302297009 Display: Scoliosis deformity of spine (disorder)
Chance_of_Kyphosis	Simple	This is a simple state for transition.	n/a
Kyphosis	Condition Onset	20% of myelomeningocele newborns have kyphosis. (9) Diagnosed at Encounter_NICU	System: SNOMED-CT Code: 414564002 Display: Kyphosis deformity of spine (disorder)
Chance_of_Survival_Day1	Simple	This is a simple state for transition. Myelomeningocele newborns' mortality rate is 6.1% on day 1. (10)	n/a
Delay_Myelomeningocele	Delay	Set the delay to 24-48 hours.	n/a
Myelomeningocele_Repair	Procedure	96% of newborns with myelomeningocele will have repair surgery within 48 hours of birth. (11) Reason: Myelomeningocele	System: SNOMED-CT Code: 42839003 Display: Repair of myelomeningocele (procedure)

State Name	Type	State Remarks	Terminology
Had_Fetal_Surgery	Condition Onset	4% of newborns had fetal surgery to repair myelomeningocele. (12) Diagnosed at Encounter_NICU	System: SNOMED-CT Code: 120991000119102 Display: History of undergoing in utero procedure while a fetus (situation)
Hydrocephalus	Condition Onset	Myelomeningocele newborns who had repair surgery have an 85% probability of having complication of hydrocephalus. Myelomeningocele newborns who had fetal surgery have a 45% chance of having hydrocephalus. (13) (14) Diagnosed at Encounter_NICU	System: SNOMED-CT Code: 230745008 Display: Hydrocephalus (disorder)
Delay_Shunt_Placement	Delay	Set the delay to 2-5 days.	n/a
ETV	Procedure	No available prevalence data for distribution. Set probability to 20% for patients who have hydrocephalus to receive combined endoscopic third ventriculostomy (ETV) and choroid plexus cauterization (CPC) procedure for treatment. Reason: Hydrocephalus	System: SNOMED-CT Code: 441763001 Display: Endoscopic third ventriculostomy (procedure)
CPC	Procedure	Patients have CPC procedure. Note that combining CPC with ETV was superior to ETV alone in infants.(15) Reason: Hydrocephalus	System: SNOMED-CT Code: 17881005 Display: Cauterization of choroid plexus (procedure)
Ventriculoperitoneal_Shunt	Procedure	No available prevalence data for distribution. Set probability to 80% for patients who have hydrocephalus to have ventriculoperitoneal shunt placed. Reason: Hydrocephalus	System: SNOMED-CT Code: 47020004 Display: Ventriculoperitoneal shunt (procedure)
Delay_Discharge	Delay	Set the delay to 3-7 days.	n/a
Chance_of_Survival_2	Simple	This is a simple state for transition. Myelomeningocele newborns mortality rate is roughly 2% on Day 2-Day 27. (10)	n/a
End_Encounter_NICU	EncounterEnd	This state ends the current encounter, Encounter_NICU. Patient is discharged from NICU.	n/a
Delay_SB_Visit	Delay	Set the delay to 2-3 months.	n/a

State Name	Type	State Remarks	Terminology
SB_Visit_1st	Encounter	This is the first spina bifida clinic visit. Reason for visit is myelomeningocele. Reason: Myelomeningocele	System: SNOMED-CT Code: 308335008 Display: Patient encounter procedure (procedure)
Urodynamic_Test	Procedure	During the first spina bifida clinic visit, all patients receive urodynamic test. Reason: Myelomeningocele	System: SNOMED-CT Code: 252895004 Display: Urodynamic studies (procedure
Renal_Ultrasound	Procedure	During the first spina bifida clinic visit, all patients receive renal ultrasound. Reason: Myelomeningocele	System: SNOMED-CT Code: 709640007 Display: Doppler ultrasonography of renal vein (procedure)
CT_Head	Procedure	During the first spina bifida clinic visit, all patients have a CT head scan. Reason: Myelomeningocele	System: SNOMED-CT Code: 303653007 Display: Computed tomography of head (procedure)
MRI_Spine	Procedure	During the first spina bifida clinic visit, all patients have an MRI for spine. Reason: Myelomeningocele	System: SNOMED-CT Code: 241645008 Display: Magnetic resonance imaging of spine (procedure)
Muscle_Testing	Procedure	During the first spina bifida clinic visit, all patients are assessed for muscle function. Reason: Myelomeningocele	System: SNOMED-CT Code: 9002005 Display: Manual testing of muscle function (procedure)
Latex_Allergy	ConditionOnset	60% of myelomeningocele patients develop latex allergy. (16) Diagnosed at SB_Visit_1st	System: SNOMED-CT Code: 300916003 Display: Allergy to latex (finding)
End_SB_Visit_1st	EncounterEnd	This ends the current encounter SB_Visit_1st.	n/a
Living_With_SB	Simple	This is a simple state for transition. If age <1 year, continues to the state Delay_3_Month. Patient will visit spina bifida clinic every 3 months. If age >= 1 year, continues to the state Living_With_SB_2.	n/a

State Name	Type	State Remarks	Terminology
Delay_3_Month	Delay	Set the delay to 3 months.	n/a
Office_Visit_Under_Age_1	Encounter	This is a spina bifida clinic visit. Reason for visit is myelomeningocele. Reason: Myelomeningocele	System: SNOMED-CT Code: 308335008 Display: Patient encounter procedure (procedure)
Condition_UTI	ConditionOnset	50% of myelomeningocele patients develop a Urinary Tract Infection (UTI). (17) Diagnosed at Office_Visit_Under_Age_1	System: SNOMED-CT Code: 68566005 Display: Urinary tract infectious disease (disorder)
End_Officie_Visit_Under_Age_1	EncounterEnd	This ends the current encounter, Office_Visit_Under_Age_1	n/a
Living_With_SB_2	Simple	This is a simple state for transition. If age <1 year, then the module loops back to the Living_With_SB state. Patient will continue to have spina bifida clinic visits every 3 months until age is >= 1 year. If age >= 1 year, then the module transitions to the Delay_6_Months state.	n/a
Delay_6_Months	Delay	Set the delay to 6 months.	n/a
SB_Visit_Under_Age_5	Encounter	This is a spina bifida clinic visit. Reason for visit is myelomeningocele. Reason: Myelomeningocele	System: SNOMED-CT Code: 308335008 Display: Patient encounter procedure (procedure)
Condition_UTI_3	ConditionOnset	50% of spina bifida patients develop UTI. (17) No prevalence data is available for patients with both UTI and pressure ulcer. Set 45% chance to have UTI alone and 5% chance of having both pressure ulcer and UTI. Diagnosed at SB_Visit_Under_Age_5 Note: UTI is modeled within a routine spina bifida encounter. Ideally, UTI would be modeled as a randomly generated condition throughout childhood rather than being tied to a series of routinely scheduled encounters. This appears to be a limitation of Synthea. Pathways of encounters for routine visits and encounters for complications cannot be modeled simultaneously in the same module.	System: SNOMED-CT Code: 68566005 Display: Urinary tract infectious disease (disorder)

State Name	Type	State Remarks	Terminology
Condition_Pressure_Ulcer_3	ConditionOnset	15.6% of spina bifida patients develop pressure ulcer. (18) Set 10.6% chance to have pressure ulcer alone. Diagnosed at SB_Visit_Under_Age_5 Note: Pressure ulcer is modeled within a routine spina bifida encounter. Ideally, pressure ulcer would be modeled as a randomly generated condition throughout childhood rather than being tied to a series of routinely scheduled encounters. This appears to be a limitation of Synthea. Pathways of encounters for routine visits and encounters for complications cannot be modeled simultaneously in the same module.	System: SNOMED-CT Code: 399912005 Display: Pressure ulcer (disorder)
Condition_Pressure_Ulcer_4	ConditionOnset	15.6% of spina bifida patients develop pressure ulcer. (18) Set 5% chance of having both pressure ulcer and UTI. Diagnosed at SB_Visit_Under_Age_5	System: SNOMED-CT Code: 399912005 Display: Pressure ulcer (disorder)
End_SB_Visit_Under_Age_5	EncounterEnd	This ends the current encounter, Office_Visit_Under_Age_5. If age <5 years, patients will have spina bifida clinic visit every 6 months while age is <5 years.. If age >=5 years, then the module transitions to the Delay_1_Year state. Patients will have spina bifida clinic visit annually. The morbidity rate for spina bifida patients < 5 years is roughly 1%. (10)	n/a
Living_With_SB_3	Simple	This is a simple state for transition. If age <5 years, then the module loops back to the Living_With_SB_2 state. Patient will continue to have spina bifida clinic visit every 6 months while age is <5 years. If age >= 5 years, then the module transitions to the Delay_1_Year state. Patients will have spina bifida clinic visit annually.	n/a
Delay_1_Year	Delay	Set the delay to 1 year.	n/a
SB_Visit_After_Age_5	Encounter	This is a spina bifida clinic visit. Reason for visit is myelomeningocele.	System: SNOMED-CT Code: 308335008 Display: Patient encounter procedure (procedure)

State Name	Type	State Remarks	Terminology
Condition_UTI_2	ConditionOnset	50% of spina bifida patients develop UTI. (17) No prevalence data is available for patients with both UTI and pressure ulcer. Set 45% chance to have UTI alone and 5% chance of having both pressure ulcer and UTI. Diagnosed at SB_Visit_After_Age_5 Note: UTI is modeled within a routine spina bifida encounter. Ideally, UTI would be modeled as a randomly generated condition throughout the childhood rather than being tied to a series of routinely scheduled encounters. This appears to be a limitation of Synthea. Pathways of encounters for routine visits and encounters for complications cannot be modeled simultaneously in the same module.	System: SNOMED-CT Code: 68566005 Display: Urinary tract infectious disease (disorder)
Condition_Pressure_Ulcer	ConditionOnset	15.6% of spina bifida patients develop pressure ulcer. (18) Set 10.6% chance of having pressure ulcer alone. Diagnosed at SB_Visit_After_Age_5 Note: Pressure ulcer is modeled within a routine spina bifida encounter. Ideally, pressure ulcer would be modeled as a randomly generated condition throughout the childhood rather than being tied to a series of routinely scheduled encounters. This appears to be a limitation of Synthea. Pathways of encounters for routine visits and encounters for complications cannot be modeled simultaneously in the same module.	System: SNOMED-CT Code: 399912005 Display: Pressure ulcer (disorder)
Condition_Pressure_Ulcer_2	ConditionOnset	15.6% of spina bifida patients develop pressure ulcer. (18) Set 5% chance of having both pressure ulcer and UTI. Diagnosed at SB_Visit_After_Age_5	System: SNOMED-CT Code: 399912005 Display: Pressure ulcer (disorder)
End_SB_Visit_After_Age_5	EncounterEnd	This ends the current encounter, Office_Visit_After_Age_5. The morbidity rate for spina bifida patients is 25%.	n/a
Living_With_SB_4	Simple	This is a simple state for transition. If age<18 years, then patients will loop back to the Delay_1_Year state. If age >=18 years, then exit the module. The morbidity rate for spina bifida patients age of 5 and above is roughly 0.5%. (10)	n/a
Death	Death	This state indicates death of the patient with spina bifida.	System: SNOMED-CT Code: 414667000 Display: Meningomyelocele (disorder)
Terminal	Terminal	Ending state of a Synthea module.	n/a

Module Parameters

Table 3: Spina Bifida Module Parameters summarizes the probabilities used to construct distributed module states where branching occurs in the module flow. A value of 1.0 indicates 100% and 0 indicates 0%.

Table 3: Spina Bifida Module Parameters

Parameter	Value	Notes and References
Probability of newborns having the 3 common types of spina bifida	1.0	n/a
1. Myelomeningocele	0.00035	(3) (4) (5)
2. Meningocele	0.0001	(3)
3. Spina Bifida Occulta	0.00025	(3)
Probability of myelomeningocele patients also having Chiari II Malformation	0.90	(6)
Probability of myelomeningocele patients having foot deformity	0.90	(7)
Probability that newborns had fetal surgery to repair open spina bifida	0.04	(12)
Probability of newborns who had fetal surgery to repair myelomeningocele developing hydrocephalus	0.45	(13)
Probability of newborns who have surgery to repair myelomeningocele developing hydrocephalus	0.85	(13)
Probability of patients developing latex allergy	0.60	(16)
Probability of patients developing UTI	0.50	(17)
Probability of patients developing pressure ulcer	0.156	(18)
Myelomeningocele mortality rate Day 1	0.061	(10)
Myelomeningocele mortality rate Day 2-Day 27	0.002	(10)
Myelomeningocele mortality rate Year under 5	0.001	(10)
Myelomeningocele mortality rate Year 5 and above	0.005	(10)

Sample Synthetic Data Results

Sample Synthea generated data results for this module are included below (see Table 4). Analysis was performed using 100,104 patients (104 deceased patients), with an age range of 0 to 18 years, generated in CSV output from Synthea. The synthetic prevalence rates matched the prevalence benchmark defined in the module for meningocele (0.007% vs. 0.010%) and myelomeningocele (0.037% vs. 0.035%). The synthetic prevalence rate for spina bifida occulta in this data set is 0.002%, whereas the benchmark prevalence defined in the module is 0.025%. This is likely due to the very low prevalence rate of these conditions; a much larger generated data set will likely reflect the prevalence rate in the generated results more accurately.

Table 4: Patients with Spina Bifida Synthetic Prevalence

Spina Bifida Type	Patients with Spina Bifida	Total Patients	Synthetic Prevalence	Benchmark Prevalence Defined in Module
Meningocele (disorder)	7	100104	0.007%	0.010%*
Meningomyelocele (disorder)	37	100104	0.037%	0.035%*
Spina bifida occulta (disorder)	2	100104	0.002%	0.025%*
Grand Total	**46**	**100104**	**0.046%**	**0.070%***

* *See* Table 3: Spina Bifida Module Parameters

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